Vol. 74 No. 7 Established 1884 July 1957

ARCHIVES OF PEDIATRICS

A MONTHLY DEVOTED TO THE

DISEASES OF INFANTS AND CHILDREN

JOHN FITCH LANDON, M.D., Editor

LEADING ARTICLES IN THIS NUMBER

Congenital Heart Malformations in Mongolism with Special Reference to Ostium Atrioventriculare Commune.

Fatma Munire Esen, M.D. 243

The Hair Sign as a Prognosticator of Allergy.

Robert Cohen, M.D. 278

E. B. TREAT & CO., Inc., *Publishers*, 45 East 17th Street, NEW YORK 3

Yearly Subscription $6.00 (Foreign $6.75); Single Copy, $1.00

COPYRIGHT, 1957, BY E. B. TREAT & CO., INC. ALL RIGHTS RESERVED

Entered as second-class matter Feb. 5, 1892, at New York, N. Y., P. O., under the Act of March 3, 1879

EVERY LISTED PEDIATRIC SPECIALIST

was questioned by an independent research organization about an article published in the Archives of Pediatrics. These specialists were asked whether they agreed with the reprint material.

Of the pediatricians who believed their experience justified an opinion, 156—81.7%—replied yes to all three points in question.

4 out of 5 Leading Pediatricians agree that CREAM OF RICE

NEW, ½ Minute Cooking Time—10 Times Faster!
NEW, Easy-Pouring Spout!

a | **gives "more available caloric energy"**
than any wheat, rye, barley, corn or oat cereal. Of the 227 pediatricians answering definitely, 192—84.6%—said yes.

b | **is "more easily digestible"**
than any other kind of cereal. Of the 248 answering definitely, 212—85.5%—said yes.

c | **gives "nutritional energy more rapidly"**
than any other kind of cereal. Of the 220 answering definitely, 178—80.9%—said yes.

In addition, Cream of Rice is

MOST HYPOALLERGENIC, TOO

As reported in the Archives of Pediatrics by Slobody, Untracht and Hertzmark, "rice . . . shows the fewest allergic reactions of any cereal checked . . . Even children potentially allergic to rice have been shown to tolerate it well when it is cooked in the presence of moisture."

WRITE FOR PROFESSIONAL SAMPLES:
GROCERY STORE PRODUCTS CO., DEPT. AP-7,
WEST CHESTER, PA.

Pops, like the Doctor said how about real apples
Beech-Nut APPLE SAUCE
Real apples coming up, Junior - natural taste, natural vitamins in Beech-Nut

TRUST BEECH-NUT...CAREFULEST BABY FEEDERS IN THE WORLD

TO CONTRIBUTORS AND CORRESPONDENTS OF

ARCHIVES OF PEDIATRICS

Subscription $6.00 a Year, in Advance **Foreign $6.75** **Single Copy, $1.00**

Editorial Communications address to JOHN F. LANDON, M.D., 120 East 75th Street, New York 21

Business Communications address to E. B. TREAT & Co., INC., 45 East 17th Street, New York 3

ORIGINAL ARTICLES, brief reports of rare and interesting cases, or new modes of treatment are solicited, but none will be considered for publication except with the distinct understanding that it is contributed exclusively to this journal. Manuscripts must be typewritten, double-spaced, and the original, not the carbon, copy submitted. The editor and publishers will not be responsible for views expressed.

ILLUSTRATIONS, as in the judgment of the editor are necessary, will be furnished free when satisfactory photographs or drawings are supplied. Photographs must be clear and distinct; drawings must be in India ink on white paper.

COPYRIGHT.—Original communications appearing in this journal are protected by copyright and can not be reproduced, either wholly or in part, without permission of the publishers.

REPRINTS of articles appearing among "Original Communications" may be ordered immediately upon receipt of galley proof by communicating direct with the publishers, E. B. Treat & Co., Inc., 45 East 17th Street, New York 3, who will supply their schedule of prices.

DISCONTINUANCES. — The publishers must be notified when a subscriber wishes his journal stopped and all arrearages must be paid. Without such notification it is assumed that a continuance is desired. Journals returned are not notice of discontinuance.

REMITTANCES should be made by check, draft, post office or express money order. If currency is sent, the letter should be registered. Stamps in amounts under one dollar are acceptable.

CHANGE OF ADDRESS NOTICE should give both the old and the new and state if change is permanent or temporary.

CONTENTS

ORIGINAL COMMUNICATIONS

Congenital Heart Malformations in Mongolism with Special Reference to Ostium Atrioventriculare Commune.

FATMA MUNIRE ESEN, M.D. 243

The Hair Sign as a Prognosticator of Allergy.

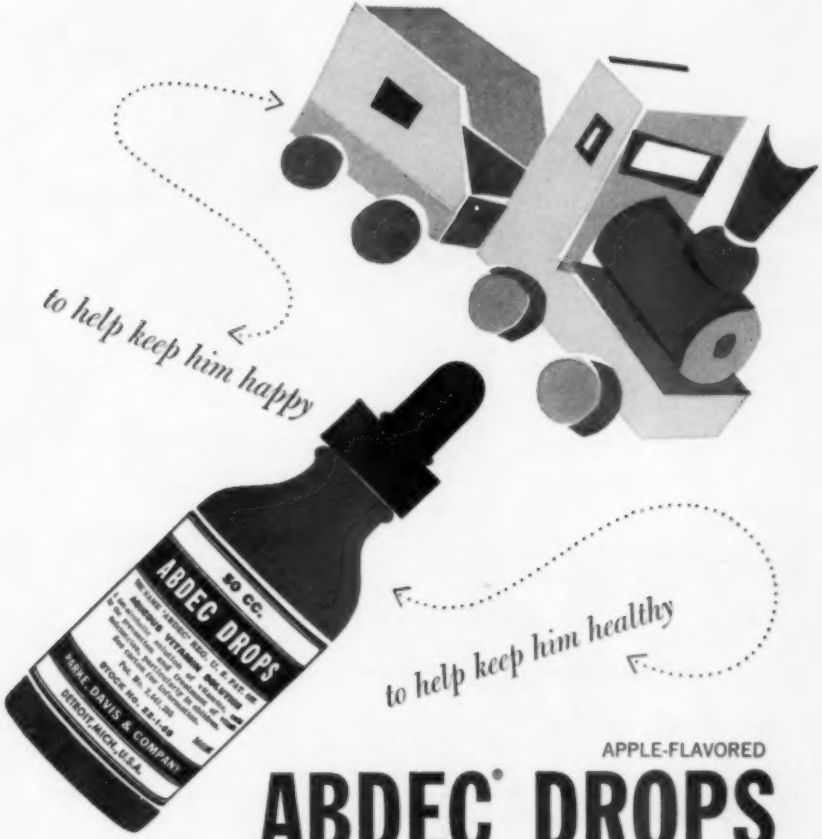
ROBERT COHEN, M.D. 278

DEPARTMENT OF ABSTRACTS

Karlin, I. W.: Stuttering. Evaluation and Treatment 280

Carter, C. H. and Maley, M. C.: Chlorpromazine Therapy in Children at the Florida Farm Colony 280

(Continued on page 4)



APPLE-FLAVORED

ABDEC® DROPS

concentrated multivitamin supplement

Each 0.6 cc. of ABDEC DROPS represents:

Vitamin A	5,000 units
Vitamin D	1,000 units
Vitamin B_1 (thiamine hydrochloride)	1 mg.
Vitamin B_2 (riboflavin)	0.4 mg.
Vitamin B_6 (pyridoxine hydrochloride)	1 mg.
Pantothenic acid (as the sodium salt)	2 mg.
Nicotinamide	5 mg.
Vitamin C (ascorbic acid)	50 mg.

MEDICAMENTA VERA

PARKE, DAVIS & COMPANY · DETROIT 32, MICHIGAN

(Continued from page 2)

Farquhar, J. W.: The Possible Influence of Hyperadrenocorticism on the Foetus of the Diabetic Woman 281

Varadi, S. and Hurworth, E.: Heinz-Body Anemia in the Newborn 282

Choremis, C.; Papadatos, C.; Gargoulas, A. and Drosos, C.: Intrathecal Hydrocortisone in the Treatment of Tuberculous Meningitis 282

Gelfand, H. M.; Fox, J. P. and Le Blanc, D. R.: Observations on Natural Poliovirus Infections in Immunized Children 283

Beach, W. B.: Psychosis in Childhood. 283

Bain, H. W.; Bowden, D. H.; Chute, A. L.; Jackson, S. H.; Sass-Kortsak, A. and Walker, N. F.: Galactosemia 284

Chalmers, T. C.; Iber, F. L. and Uzman, L. L.: Hepatolenticular Degeneration (Wilson's Disease) as a Form of Idiopathic Cirrhosis 284

AS THEY FIND IT –

"PRACTICAL"

"The very concise and brief clinical reports that appear in the ARCHIVES each month have been very helpful to me, and *I have found more practical suggestions in them than most of the long and drawn out scientific articles that appear in other journals.*"

"I want to tell you that after subscribing for about 16 years, *I get more practical pediatrics from your journal than from either of the other pediatric journals to which I subscribe.*"

"Because it is *concise and progressive* I consider ARCHIVES OF PEDIATRICS my favorite among pediatric journals. I always look forward with pleasure to the receipt of each number and *glean much of value from it.*"

"The only pediatric journal now on file there (Ellis Hospital, Schenectady) is the American Journal of Diseases of Children and I feel that *yours will be of more practical value to the general practitioners.*"

(*Names on Application*)

ARCHIVES OF PEDIATRICS

45 East 17th St., New York

one application of

DESITIN® OINTMENT

helps protect the infant's skin against

diaper rash (ammoniacal dermatitis) • **irritation** • **excoriation**

- tubes of 1 oz., 2 oz., 4 oz.
- 1 lb. jars.

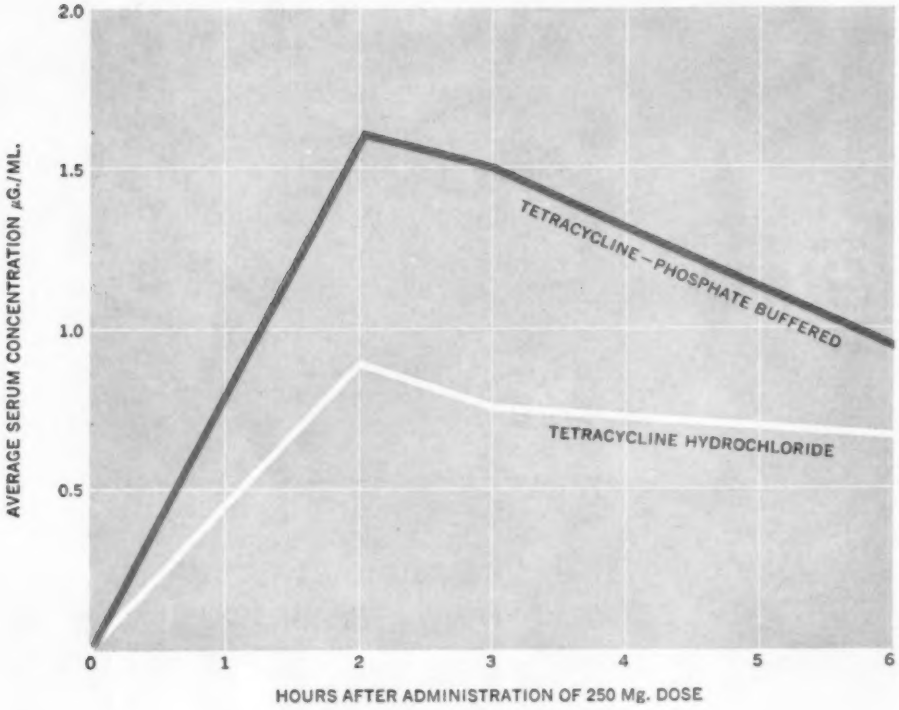
DESITIN OINTMENT covers the infant's skin with a soothing, protective, healing coating which is largely impervious to and helps guard against irritation, rash, and maceration caused by urine, excrement, perspiration and secretions. This preventive action of Desitin Ointment *persists all through the night*...when baby is particularly vulnerable to painful skin excoriations.

Nonsensitizing, nonirritant Desitin Ointment.. **rich in cod liver oil** ....successfully used on millions of infants for over 30 years.

for **samples** *and literature please write....*

DESITIN CHEMICAL COMPANY Providence, R.I.

1. Grayzel, H. G., Heimer, C. B., and Grayzel, R. W.: New York St. J. Med. 53:2233, 1953. **2.** Heimer, C. B., Grayzel, H. G., and Kramer, B.: Archives of Pediatrics 68:382, 1951. **3.** Behrman, H. T., Combes, F. C., Bobroff, A., and Leviticus, R.: Ind. Med. & Surgery 18:512, 1949. **4.** Turell, R.: New York St. J. Med. 50:2282, 1950. **5.** Marks, M. M.: Missouri Med. 52:187, 1955.



tetracycline activated for higher, faster levels

TETRABON® V

TETRACYCLINE-PHOSPHATE BUFFERED — HOMOGENIZED SYRUP

activated for higher, faster tetracycline blood levels . . . potent, prompt, prolonged control of tetracycline-susceptible infections . . . prescription adherence assured . . . no reconstitution required

new,
orange-flavored
delight in
broad-spectrum
therapy

Supplied:
Each 5 cc. teaspoonful of Tetrabon V contains tetracycline equivalent to 125 mg. tetracycline hydrochloride, phosphate buffered, in an orange-flavored, orange-colored, ready-to-use, homogenized suspension. Bottles of 2 oz. and 1 pint.

Pfizer

PFIZER LABORATORIES, Brooklyn 6, N. Y.
Division, Chas. Pfizer & Co., Inc.,

ARCHIVES OF PEDIATRICS

Vol. 74 JULY 1957 No. 7

JOHN FITCH LANDON, M.D., Editor

EDITORIAL BOARD

HAROLD R. MIXSELL, M.D., New York PHILIP M. STIMSON, M.D., New York
FREDK. H. WILKE, M.D., New York JOHN ZAHORSKY, M.D., St. Louis

P. W. BRAESTRUP, M.D., Hellrup, Denmark
RONALD N. MacLEAN, M.D., Buenos Aires, Argentina
Y. NISHIZAWA, M.D., Osaka, Japan
ROALD RINVIK M.D., Oslo, Norway

CONGENITAL HEART MALFORMATIONS IN MONGOLISM WITH SPECIAL REFERENCE TO OSTIUM ATRIOVENTRICULARE COMMUNE*

FATMA MUNIRE ESEN, M.D.**
Wrentham, Mass.

This paper is a report on our findings on congenital heart malformations in autopsies of mongoloids.

The hearts of mongoloid children frequently show congenital abnormalities such as: ventricular septal defects, atrial septal defects, patent ductus arteriosus, and an ostium atrioventriculare commune. In general, the cardiac septal defects appear in from one-half to two-thirds of mongoloid children. According to Benda, 75 per cent of newborn mongoloids who die in the first and the second year of life show congenital heart malformation; of those who survive, about 35 per cent. Other authors give the following figures: Scheer, 43.5 per cent; Orel, 16.2 per cent; Illing, 7.4 per cent; Kaiser and Schmid, 22.5 per cent; Geyer, 20.6 per cent.

Evans stated that nearly half of a series of mongoloids dying in the first five years of life showed congenital heart malformations. He found twenty-eight heart malformations in sixty-three autopsies of mongoloids. The ventricular and atrial septal defects each accounted for over one-third, and patent ductus arteriosus

*From the Wrentham State School and Wallace Research Laboratory, Wrentham, Mass.
**Pathologist, Wrentham State School, Wrentham, Mass.

for one-sixth. Persistent ostium atrioventriculare commune and pulmonary stenosis each occurred in four patients. This was compared with a control group of heart malformations without mongolism. In this group, he found transposition of the great vessels, coarctation and truncus arteriosus which he had not seen in mongoloids. Atrial septal defects and pulmonary stenosis were more common in the mongoloids than in the control group. He described the differences between the lesions in these two series by the absence in the mongoloids of defects due to malrotation of the bulbus cordis and the absence of a common atrioventricular opening in the other children. The atrial septal defects were more common in mongoloids than in the other children. Ventricular septal defects were equal in the two groups, while a patent ductus arteriosus was less common in the mongoloids.

Kaiser and Schmid described the atrial septal defects, subaortic ventricular septal defects, patent ductus arteriosus as a milder form of congenital heart malformations in mongolism. The most severe form was the persistent ostium atrioventriculare commune, which occurred in seven cases.

Between February 1951, and April 17, 1957, one hundred and forty-nine autopsies were performed on mentally retarded children at the Wrentham State School.

Thirty-one of them were typical mongoloids. Twelve out of the thirty-one showed congenital heart malformations. The milder forms were atrial septal defects, ventricular septal defects, patent ductus arteriosus. The severe and more complicated forms were an ostium atrioventriculare commune which was observed in seven out of twelve cases with congenital heart malformations.

CASE REPORTS

I. *Case 1.* S.H.J., white male, born December 23, 1947 at full term by normal delivery. First pregnancy. Mother was 21 years of age at birth of patient and her health was good during pregnancy. There were two younger, normal siblings. Parents are of normal intelligence and in good health. Mongolism was diagnosed at birth and institutionalization was recommended. The baby was admitted to the Wrentham State School on April 1, 1950 at the age of 2 years and 3 months. The *physical examination* revealed a 95 cm. tall typical mongoloid infant who weighed 1400 gms. The head measured 45 cm. in circumference, 12½ cm. in width, and 14½ cm. in length. He had curved little fingers with small middle phalanges, large distance between first and second toes, atypical

four finger (Simian) line, flattened helices, slanting palpebral fissures, epicanthal folds, blue eyes, small nose with broad nasal bridge, convergent strabismus, large fissured tongue, marked muscular hypotonicity, pulse 150 per minute. First teeth appeared at one year of age. He did not talk nor walk. The *psychological examination* revealed a mental age (M.A.) of 4 months and an I.Q. of .17, (Kuhlmann-Binet).

The erythrocytes numbered 4,810,000 per cubic millimeter with 65 per cent polymorphonuclear, 34 per cent lymphocytes, 1 per cent Turk cell. The erythrocytes and platelets were normal. The Hinton test was negative.

Course: The baby had a good appetite and was spoon fed; slept well, but took very little interest in his surroundings. He moved around the crib but was not able to pull himself up. At the age of 2 years and 9 months, he developed frequent upper respiratory infections and became a feeding problem. He later vomited often and lost weight. At the age of 3 years, 1 month and 7 days, he developed a severe bronchitis with cyanosis and irregular pulse. Oxygen was administered and antibiotic therapy started, but without success. The child died on the same day.

Autopsy Findings: The pericardium contained a few drops of clear, straw colored fluid. The heart was enlarged weighing 65½ gm., (normal weight, 57½ gm.). The interventricular septum showed a defect. The wall of the left ventricle measured 4 mm. in thickness, and the wall of the right ventricle, 1 mm. The ductus arteriosus and foramen ovale were closed. The coronary arteries were normal and in normal position. *Other findings at autopsy:* Acute catarrhal, membranous, fibrinous pharyngitis; catarrhal tracheo-bronchitis; bronchopneumonia; extensive pial edema; fatty degeneration of the liver; relatively fresh invagination of the small intestine.

Diagnosis: Mongolism; interventricular septal defect.

II. *Case 2.* C.M., white female, born October 11, 1950 at full term by normal delivery. Second pregnancy. Mother was 33 years of age at birth of the patient. Her health was good during pregnancy, except for extreme nervousness at one and a half months of pregnancy due to a sudden operation upon her son for a strangulated hernia. The family history is negative. Parents are healthy and of normal intelligence. The first pregnancy resulted

in a normal child. The second baby was admitted to the Wrentham State School at two months of age on December 16, 1950. The *physical examination* revealed a girl, 57 cm. tall weighing 5000 gms. She showed typical mongoloid features with flattened helices, slanting palpebral fissures, marked epicanthal folds, small nose with broad nasal bridge, a large tongue, short extremities, hands and fingers, as well as feet and toes, four finger (Simian) lines in both palms. The head measured 37 cm. in circumference, 12.4 cm. in length, and 10.2 cm. in width. The frontal fontanelle was open 5 cm. by 4 cm. The other fontanelles were palpable. The abdomen was mildly protruding. There was a marked convergent strabismus. During the examination the baby showed mild cyanosis around the mouth, on palms, and soles of feet, when she cried. The respiration was 70; pulse 170. The heart was not enlarged, but there was a very loud, machinery-like systolic murmur at the second intercostal space at left border of sternum. The *psychological examination* on admission showed an M.A. of 18 days, and an I.Q. of .26, (Kuhlmann-Binet).

The erythrocytes numbered 3,940,000 per cubic millimeter and the hemoglobin was 13.3 gm., (Fisher). The leukocytes numbered 7,200 per cubic millimeter with 40 per cent polymorphonuclear, 41 per cent lymphocytes, 15 per cent monocytes, 4 per cent plasma cells. The erythrocytes showed moderate achromia, frequent target cells, moderate anisocytosis, (micro). The platelets were normal. The Hinton test was negative. The roentgenogram showed enlargement of the right ventricle.

Course: The baby was rather apathic, listless, developed cyanosis when she cried and when fed. At eleven months of age she started to have frequent, mild attacks of cyanosis, mostly combined with respiratory infections, which continued until death. She died suddenly at the age of 1 year, 3 months, 3 days of severe heart failure.

Autopsy Findings: The pericardium contained 15 cc. clear, straw colored fluid. There were scattered fibrin deposits on the surface of the right ventricle. The heart was markedly enlarged weighing 130 gms., (normal weight, 46½ gms.). The right ventricle showed marked hypertrophy. The lower portion of the atrial septum showed a large defect which was combined with the large ventricular septal defect. The tricuspid and mitral orifices were not separated, and one large atrioventricular orifice was present. There was only one atrioventricular valve, because of

fusion of the posterior cusp of the tricuspid valve with the posterior half of the anterior leaflet of the mitral valve, and the fusion of the anterior half of the mitral cusp with the medial cusp of the tricuspid valve. The membrane which occluded the foramen ovale showed a small defect. The ductus arteriosus was represented by a ligamentous cord. The wall of the left ventricle measured 5 mm. in thickness and the wall of the right ventricle, 7 mm. The pulmonary artery showed mild stenosis. The ascending aorta was markedly dilated. The coronary arteries were in normal position. *Other findings at autopsy:* Marked pulmonary edema, marked fatty degeneration of the liver; the remaining organs showed marked hyperemia.

Diagnosis: Mongolism; ostium atrioventriculare commune; right ventricular hypertrophy; stenosis of the pulmonary orifice and pulmonary trunk; dilatation of the ascending aorta; dilatation of left ventricle; patent foramen ovale.

III. *Case 3.* S.L., white female, born December 29, 1951 at full term by normal delivery. Fourth pregnancy. Mother was 40 years of age at birth of the patient and was Rh negative. In the second and third month of pregnancy she had vaginal bleeding due to threatened abortion. Mother's health was good for the rest of the pregnancy. Mongolism was diagnosed at birth. The baby was severely jaundiced, but she ate vigorously and had no postnatal distress while in the hospital. Parents are healthy and of normal intelligence. First pregnancy resulted in a pair of premature twins following a toxemic prenatal course, (pre-eclampsia and hydramnion). One twin was normal, the other was a monstrosity with marked edema of the body who died one hour after birth. Result of the second pregnancy was a normal child. The third pregnancy was terminated by miscarriage at three months due to threatened abortion in November 1950. The father's nephew (not mongoloid) was a mentally retarded patient at Wrentham State School. The baby was admitted to the Wrentham State School at the age of twenty days on January 18, 1952. The *physical examination* revealed a well nourished mongoloid infant, who was 49½ cm. tall and weighed 3500 gms. She showed typical mongoloid features with slanting palpebral fissures, flattened helices, epicanthal folds, small nose with broad nasal bridge, large tongue, hypotonic muscles, small umbilical hernia. Head measured 43½ cm. in circumference, 11.9 cm. in length, and 9½ cm. in width. Respiration was 36, pulse 128. The frontal fontanelle was largely

open. Examination of the heart revealed no murmur. The *psychological examination* showed an M.A. of 23 days and an I.Q. of 100.

The erythrocytes numbered 3,830,000 per cubic millimeter, and the hemoglobin was 13.3 gms., (Fisher). The leukocytes numbered 10,950 per cubic millimeter with 38 per cent polymorphonuclear, 43 per cent lymphocytes, ½ per cent plasma cell, 18½ per cent monocytes. The erythrocytes showed slight achromia, slight anisocytosis, (macro), slight polychromasia. The blood platelets were normal. The Hinton test was negative.

Course: The baby was well to the age of four months. Before this date she ate and slept well and seemed to be aware of all that went on about her. Then she developed bronchopneumonia with marked, recurrent dyspnea and cyanosis and was treated with antibiotics without success. Coramine and oxygen were administered. In spite of constant hydration, continued oxygen and antibiotics, the baby died at four months of age.

Autopsy Findings: The pericardium contained 3 cc. clear, straw colored fluid. The heart was large and weighed 60 gms., (normal weight for this age, 27 gms.). The atrial septum showed two defects, one referred to the foramen ovale, the other, which was much larger, extended on the lower half of the atrial septum to the border of the mitral and tricuspid valves. The mitral and tricuspid valves were narrow. Both atria showed dilatation, especially the right. The left ventricle was rather hypoplastic, while the right ventricle was markedly hypertrophic and dilated. The wall of the left ventricle measured 5 mm. in thickness and the wall of right ventricle 8 mm. The ductus arteriosus was represented by a ligamentous cord. The coronary arteries were normal and in normal position. *Other findings at autopsy:* Extensive, partially hemorrhagic bronchopneumonia, (aspiration pneumonia); congenital malformation of middle lobe of right lung; presence of small fissure; hypoplasia of the adrenal and pituitary glands; marked diffuse swelling and numerous hemorrhagic erosions of the intestinal mucosa.

Diagnosis: Mongolism; persistent ostium primum; hypertrophy and dilatation of the right ventricle; dilatation of both atria; hypoplasia of left ventricle; patent foramen ovale.

IV. *Case 6.* C.J.R., white male, born May 26, 1951 at full term by normal delivery. Fourth pregnancy. Mother was 40 years of age at birth of the patient and suffered from a rheumatic heart

disease during pregnancy and was emotionally disturbed due to the care of her other mongoloid child. First and second pregnancies resulted in normal children. Third pregnancy with twins in 1948, of which one died at birth, an infant girl, (not known if a mongoloid), and the other was a mongoloid and is at Wrentham State School. Parents are of normal intelligence. Following birth, the baby was not shown to the mother and remained in the hospital until admission to the Wrentham State School on July 9, 1951 at the age of one month.

The *physical examination* showed a tiny mongoloid boy, 56 cm. tall and weighing 3000 gms. He had typical mongoloid features with slanting palpebral fissures, flattened helices, small nose with broad nasal bridge, large tongue, short extremities, hands and fingers as well as feet and toes, with large first toes; protruding abdomen. The little fingers were not curved. The head measured 35 cm. in circumference, 11 cm. in length and 9½ cm. in width. The fontanelles and sutures were broadly open. The pulse was irregular, it dropped suddenly from 140 to 70 and rose when he cried. There were no heart murmurs present and no cyanosis was observed, but there was an increased pulsation visible at the middle of the heart dullness. The *psychological examination* revealed an M.A. of 1 month and an I.Q. of .67, (Cattell Infant Scale).

Examination of the peripheral blood revealed 5,330,000 per cubic millimeter erythrocytes. The leukocyte count was 8,050 per cubic millimeter with 49 per cent polymorphonuclear, 39½ per cent lymphocytes, 11 per cent monocytes, ½ per cent plasma cells. The erythrocytes showed slight to moderate anisocytosis, (macro), slight polychromasia. The platelets were normal. The Hinton test was negative.

Course: The patient was well to the age of 7 months, then he developed respiratory infections which continued intermittently until his death. The x-ray examination showed signs of pneumonia. In spite of antibiotic treatment, he continued to have density in the lung field and had the appearance of a very sick baby. After initial response to antibiotic treatment his temperature rose again to 106; the baby became restless, cyanotic and in spite of oxygen therapy he died at the age of 11 months, 23 days. Blood culture was negative.

Autopsy Findings: The pericardium contained a few drops of clear straw colored fluid. The heart weighed 50 gms., (normal

weight, 42 gms.). There was pericardial thickening of the anterior and posterior surface of the right ventricle. The atrial septum showed a large defect of the lower portion which combined with the defect of the interventricular septum. The anterior mitral cusp was united with the anterior cusp of the tricuspid valve; the posterior mitral cusp and posterior tricuspid cusp were united as well. The tricuspid and mitral orifices were not separated and one large atrioventricular orifice was present, with a common atrioventricular valve. The left ventricle was intensely dilated and showed thrombi between the trabecular muscles. The pulmonary trunk showed a marked diffuse dilatation; also the right ventricle was markedly dilated. The aorta was relatively narrow. The coronary arteries were normal and in normal position. The foramen ovale showed moderate opening which measured 4 mm. in diameter. The ductus arteriosus was widely open. The wall of both ventricles measured 8 mm. in thickness. *Other findings at autopsy:* Lobular pneumonic infiltration of both lungs; marked congestional swelling and induration of the liver and spleen; fresh selective necrosis of the cortex of the right kidney; fresh embolic-thrombotic occlusion of one of the renal arteries of right kidney; myocarditis.

Diagnosis: Mongolism; ostium atrioventriculare commune; patent ductus arteriosus; marked dilatation of left ventricle; hypertrophy of right ventricle; patent foramen ovale.

V. *Case 9.* B.L.P., white male, born April 7, 1952 two weeks before expected date by normal delivery. The cord was around the neck and the baby was pale. Birth weight 5 pounds, 1 ounce. He was in an oxygen tent for 1½ days. Second pregnancy. Mother was 27 years of age at birth of patient and had rheumatic heart disease since the age of 14 years. She had three episodes of vaginal bleeding during the first 2½ months of pregnancy. At 17 years of age mother was treated with a thyroid preparation because of obesity but no thyroid disease was found. The family history is negative on both sides. Result of first pregnancy was a normal child. Parents are of normal intelligence. The baby was a feeding problem, because of poor sucking reflexes. At six weeks of age the parents noticed a peculiarity and they brought the child at the eighth week to the Children's Medical Center. There, Mongolism was diagnosed. At the age of three months, the baby developed an upper respiratory infection which occurred intermittently until his death. The baby was admitted to the

Wrentham State School on November 13, 1952 at the age of 7 months and 6 days. The ***physical examination*** revealed a typical mongoloid baby who was 58½ cm. tall. There was a marked systolic murmur over the precardium. The irises showed heterochromia. The head measured 39½ cm. in circumference. The frontal and occipital fontanelles were largely open. The *psychological examination* revealed an M.A. of 2½ months and an I.Q. of .35. (estimated).

The x-ray examination of the heart showed marked enlargement with pulmonary peribronchial infiltration.

Course: A few hours after admission, the baby became apneic with a marked cyanosis. Coramine, aspiration by catheter and mouth suction, artificial respiration was given. The baby began to breathe again irregularly at first and after a few minutes satisfactory respiration started on oxygen administration; this was maintained for about 40 minutes. While under observation, baby's heart stopped beating and the baby died on admission day at 7 months, 6 days of age.

Autopsy Findings: The pericardium contained 50 cc. of clear straw colored fluid. The heart was moderately enlarged, weighing 55 gm. (normal weight, 34 gm.). It measured 65 mm. in vertical diameter, 60 mm. in transverse diameter and 75 mm. in oblique diameter. The aorta measured 30 mm. in circumference at the valve. The aortic valve measured 28 mm. in circumference, and was normal except for moderate fenestration of the cusps. The coronary arteries were normal and in normal position, but the right coronary artery showed two orifices; one was small and the other large. The ascending aorta was dilated. The pulmonary artery measured 35 mm. in circumference at the valve. The pulmonary valve measured 35 mm. in circumference and the cusps showed moderate fenestration with slight thickening. The pulmonary trunk and pulmonary orifice showed marked aneurysm. The right atrium was large measuring 85 mm. in transverse diameter and 20 mm. in sagittal diameter, (including the auricular appendage). The venae cavae and coronary veins were normal. The pulmonary conus and right ventricle was heavily trabeculated. The orifices of coronary sinus was enlarged measuring 5 mm. The lower portion of interatrial septum showed a large defect which combined with the ventricular septal defect. The left atrium measured 55 mm. in transverse diameter, and 20 mm. in sagittal diameter. The pulmonary veins enter in their usual position. The

ventricle was divided by an incomplete septum, which showed 5 mm. thickening and extended up for a distance of 25 mm. from the apex and left a large, round defect superiorly, measuring 25 by 25 mm. The common atrioventricular orifice measured 95 mm. in circumference. The common atrioventricular valve measured 90 mm. in circumference and was composed of four leaflets. Two of these were connected with the pulmonary conus. The tendons of these two leaflets were fastened to the anterior wall of the pulmonary conus. The other two leaflets were large, located in the middle of the common ventricle, (medial leaflet). Some of the tendons of these end in the papillary muscles of the left ventricle, and the others are fastened on the interventricular septum. The wall of both ventricle measured 7 mm. in thickness. The foramen ovale measured 5 mm. in diameter. The circumference of the ductus arteriosus was 6 mm. *Other findings at autopsy:* Extensive bronchopneumonia; hypoplasia of adrenal glands; swelling of the liver and spleen, myocarditis.

Diagnosis: Mongolism; ostium atrioventriculare commune; patent ductus arteriosus; patent foramen ovale; aneurysm of pulmonary orifice and pulmonary trunk; hypertrophy of the right ventricle; dilatation of the ascending aorta.

VI. *Case 12.* M.M., white female, born June 23, 1954 at full term by low forceps delivery. First pregnancy. Birth weight 4 pounds, 7 ounces. Mother was 27 years of age at birth of patient; her health was good in general except that she had a severe infection with sore throat during the second month of pregnancy. No other children in the family. Mongolism was diagnosed at birth and institutionalization was recommended. The baby was admitted to the Wrentham State School at the age of one month, twelve days. The *physical examination* revealed a tiny, quiet mongoloid infant who weighed 3300 gms. and was 51 cm. tall. She showed typical mongoloid features with flattened helices, slanting palpebral fissures, epicanthal folds, a small nose with a broad nasal bridge, a large fissured tongue, short extremities, hands and fingers as well as feet and toes; four finger (Simian) line on left hand. There was a gap between the first and second toes, and skin crevices crossed the foot soles from the gap. The head measured 34 cm. in circumference, 11.2 mm. in length and 8.7 cm. in width. The eyes were blue with heterochromia. The baby breathed with deep depression of the diaphragmatic region. All six fontanelles were open. The *psychological examination* on

admission revealed a M.A. of 1 month with an I.Q. of .33, (Cattell Infant Scale).

The erythrocytes numbered 4,070,000 per cubic millimeter, and hemoglobin, 15.4 gms. (Fisher). The leukocytes numbered 9,300 per cubic millimeter with 59 per cent polymorphonuclear, 7 per cent transitionales, 34 per cent lymphocytes. Blood platelets were normal. The erythrocytes showed slight achromia, slight anisocytosis, slight polychromasia and rare target cells. The Hinton test was negative.

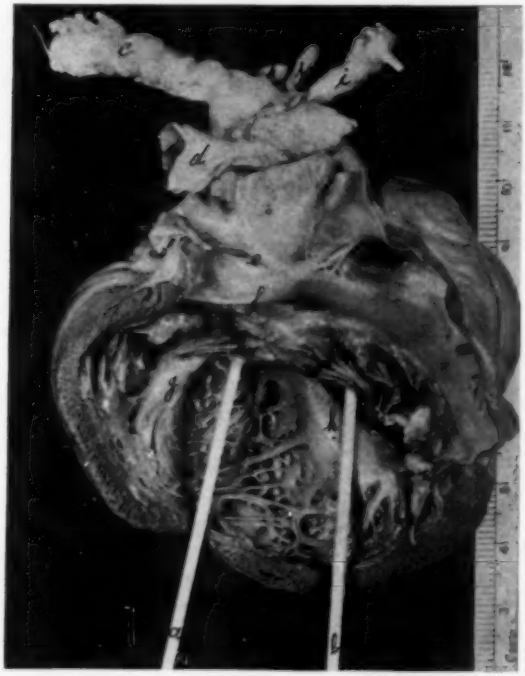


Fig. 1. Case 12. Heart of a 5½ month old mongoloid girl. Ostium atrioventriculare commune: (a) leading to aorta; (b) leading to pulmonary artery; (c) aorta; (d) pulmonary artery; (e) foramen ovale; (f) remnant of atrial septum; (g) papillary muscle; (h) remnant of ventricular septum; (i) innominate artery; (j) left carotid artery; (k) left subclavian artery.

Course: The infant took her feedings well, but showed little weight gain. At five months of age she developed cyanosis, but there were no heart murmurs. The liver was not enlarged. An

electrocardiogram was read by Dr. Conger Williams as follows: "Complexes somewhat obscured by movement; T-1, T-2, upright; moderate right axis deviation; chest shows mostly positive complexes V-1, V-2, diphasic complexes V-5. This tracing is consistent with right ventricular hypertrophy." Shortly after the first cyanotic spell, the baby developed a severe cyanosis and died at the age of 5 months and 13 days.

Autopsy Findings: The pericardium contained about 70 cc. of clear straw colored fluid. There was a thin, fibrinous infiltration over the pericardial surface. The aorta and the pulmonary artery were in normal position. Aortic and pulmonary valves were intact. The heart was enlarged weighing 57 gms., (normal weight, 30 gms.). The shape of the heart was round and had no definite apex. It measured 75 mm. in vertical diameter, 55 mm. in transverse diameter, 80 mm. in oblique diameter. The aorta measured 35 mm. in circumference at valve. The aortic valve measured 35 mm. in circumference and also had normal cusps. The pulmonary valve measured 35 mm. in circumference. The pulmonary artery measured 30 mm. in circumference at the valve. Both the aortic and pulmonary cusps showed moderate fenestration. The aorta and pulmonary artery revealed moderate aneurysm right above the valves. The coronaries were normal and in normal position. The right atrium was large measuring 80 mm. in transverse diameter, and 30 mm. in sagittal diameter (including the auricular appendage). The venae cavae and coronary veins were normal. The left atrium was very small measuring 25 mm. in transverse diameter, and 20 mm. in sagittal diameter. The pulmonary veins enter in their usual position. The lower half of the interatrial septum showed a marked defect which combined with an interventricular septal defect. The interventricular septum was completely absent, and only one large common ventricle present. A heavy ledge, over the base of the common ventricle was observed which demarcates a small chamber in the right anterior portion of the heart. This corresponds to the pulmonary conus, from which the pulmonary artery emerged. The wall of the conus pulmonalis measured 10 mm. in thickness. The wall of the common ventricle measured 6 mm. in thickness. The common atrioventricular orifice measured 80 mm. in circumference. The common atrioventricular valve measured 70 mm. in circumference and was moderately thick. The common atrioventricular valve was composed of three leaflets; two of them were connected with the conus pulmonalis. The ten-

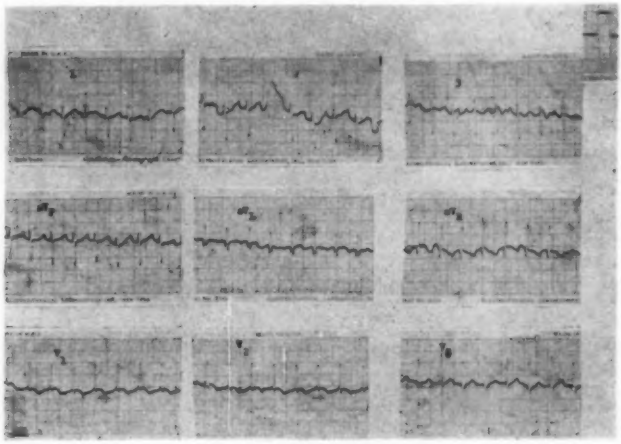
dons of these fastened to the anterior wall of the pulmonary conus. The other was rather large and laid on the middle of the common ventricle, (medial leaflet). Some of the tendons of this leaflet ended on a papillary muscle which resembles a papillary muscle of the left ventricle. The foramen oval measured 3 mm. in diameter. The circumference of the ductus arteriosus was 10 mm. *Other findings at autopsy:* Marked fatty degeneration of the liver; edema of the lungs; pia and brain.

Diagnosis: Mongolism; absence of interventricular septum; ostium atrioventriculare commune; patent ductus arteriosus; marked atresia of left atrium; and auricle; well developed pulmonary conus; marked dilatation of the right atrium and auricle; patent foramen ovale.

VII. *Case 13.* W.L.J., white female, born May 7, 1953 at full term by caesarian delivery because of mother's flattened pelvis. Third pregnancy. First two children were also delivered by caesarian section. Birth weight was 5 pounds, 8 ounces. Mother was 36 years of age at birth of patient, and her health was poor during pregnancy due to a rheumatic heart disease with mitral stenosis. Total weight gain during pregnancy was 33 pounds. Mother was on a salt free diet. Mongolism was noticed at birth and institutionalization was recommended. Mother had thyroid disease during first and second pregnancies which resulted in normal children. There were no miscarriages or stillbirths. The baby was admitted to the Wrentham State School on May 22, 1953 at the age of two weeks. The *physical examination* revealed a poorly developed mongoloid baby who was 48 cm. tall and weighed 2700 gms. The muscle tone was poor with overstretching of the joints. The shape of the head was brachycephalic with the circumference of 32½ cm., length 10½ cm., and width 9.2 cm. The sagittal suture, frontal and occipital fontanelles were broadly open. Epicanthal folds were present; the nose was small with a broad nasal bridge; the ears were small with typical flat helices. The extremities were short, hands and fingers as well as feet and toes. The little fingers showed marked curvature with a small middle phalanx. There was a large gap between the first and second toes. The abdomen was protruding. The reflexes were lively. Pulse was 160; respiration 50. The *psychological examination* at six months of age revealed a M.A. of 3.8 months, and an I.Q. of .62, (Cattell Infant Scale).

The erythrocytes numbered 5,170,000 per cubic millimeter, and the hemoglobin 14 gm. Almost all erythrocytes appeared macro in size. The leukocytes numbered 8,500 per cubic millimeter, with 56½ per cent polymorphonuclear, 7½ per cent monocytes, 3½ per cent plasma cells, 32½ per cent lymphocytes. The blood platelets were normal. The Hinton test was negative. The roentgenogram of the chest revealed marked cardiac enlargement.

Course: The baby was a very quiet, inactive infant who has shown little weight gain despite good appetite. At one month of age, she developed cyanosis with cardiomegaly and a harsh, low-pitched, apical systolic murmur. The liver was felt 1 cm. below the right costal margin. From time of admission to her death, she had often severe cyanosis and apnea with the aspiration of food. On one occasion, apnea lasted 3 minutes. An electrocardiogram was done and read by Dr. Congor Williams as follows:



Case 13—ECG.

"Sinus tachycardia at an average rate of 150; T-1, T-2, upright; T-3, low; moderate right axis deviation; chest leads show positive complexes V-1, V-2; diphasic complexes V-6." The baby had frequent upper respiratory infections with a high temperature and cyanosis. At the age of one year, 8 months, 26 days she developed severe cyanosis and died.

Autopsy Findings: The pericardium contained 50 cc. of clear, straw colored fluid. The aorta, pulmonary arteries were normal.

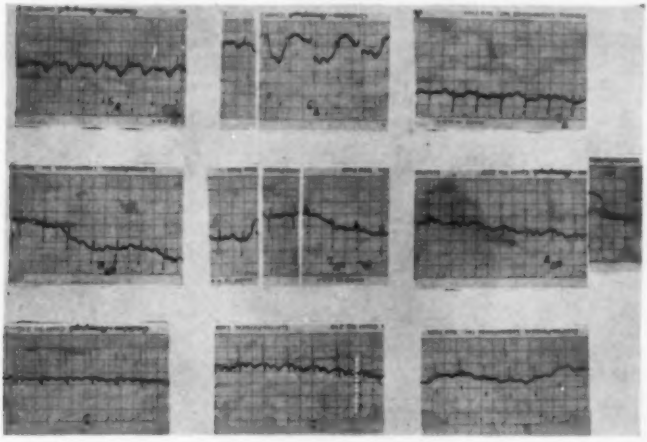
The gross examination of the heart showed a large, strong right ventricle and a small left ventricle. The heart was enlarged weighing 90 gms., (normal weight for this age, 56 gms.). It presented a vertical diameter of 80 mm., a transverse diameter of 60 mm., and an oblique diameter of 75 mm. The aortic valve was 30 mm. in circumference and showed normal cusps. The aorta measured 30 mm. in circumference at the valve. The pulmonary valve measured 35 mm. in circumference, and the pulmonary artery measured 45 mm. in circumference at the valve and showed marked dilatation. Both the aortic and pulmonary cusps showed moderate fenestration with a thick nodulus of Arantius. The coronary arteries were normal and in normal position. The right atrium was very large measuring 85 mm. in transverse diameter, and 40 mm. in sagittal diameter, (including the auricular appendage). The venae cavae and coronary veins were normal. The coronary sinus was enlarged, measuring 12 mm. in diameter. The left atrium was small measuring 30 mm. in transverse diameter, 20 mm. in sagittal diameter, and received four pulmonary veins in their normal position. There was a large defect in the lower portion of interatrial septum, which combined with the defect of interventricular septum, leaving one large atrioventricular orifice. The ventricle was divided by an incomplete septum, which showed 10 mm. in thickening and extended up for a distance of 28 mm. from the apex and left a large, round defect superiorly, measuring 35 mm. by 25 mm. The common atrioventricular orifice measured 90 mm. in circumference. The common atrioventricular valve measured 85 mm. in circumference, showed marked fibrous thickening and was composed of three leaflets. The half of the tendons of these leaflets were connected with the anterior and posterior wall of the left ventricle and the other half was fastened on to the papillar muscle of the anterior wall of the right ventricle. The wall of the right ventricle measured 11 mm. in thickness and was heavily trabeculated. The wall of the left ventricle measured 10 mm. The coronary arteries had the usual position, but the right coronary showed three orifices. The foramen ovale was largely open, measuring 10 mm. in diameter. The subclavian artery emerged from the thoracic aorta distal to the entry of the ductus arteriosus and coursed behind the esophagus to reach the right axilla. Ductus arteriosus was represented by a ligamentous cord. *Other findings at autopsy:* Partially hemorrhagic bronchopneumonia, (aspiration pneumonia); marked hyperemia and induration of the liver; congestion of the pia and swelling of the brain.

Diagnosis: Mongolism, ostium atrioventriculare commune; hypertrophy and dilatation of the right atrium and right ventricle; hypoplasia of the left atrium and auricle; patent foramen ovale.

VIII. *Case 22.* I.A.C., white male, born January 31, 1955 at 8 months by normal delivery. Third pregnancy. Mother was 32 years of age at birth of patient. Birth weight was 5 pounds, 15 ounces. During the first trimester of pregnancy, mother had vaginal bleeding for a few weeks and was hospitalized with a diagnosis of threatened abortion. Result of first pregnancy was a healthy girl of normal intelligence. The second pregnancy ended with miscarriage, shortly before she became pregnant with this patient. Parents are healthy and of normal intelligence. The family history is negative on both sides. Mongolism was diagnosed at birth. He remained in the hospital until admitted to the Wrentham State School on February 18, 1955 at the age of eighteen days. The *physical examination* showed a mongoloid infant who was 48 cm. tall and weighed 2949 gms. He had typical mongoloid features with slanting palpebral fissures, flattened helices, epicanthal folds, small nose with a broad nasal bridge, a large tongue, hypotonic muscles, four finger (Simian) lines on both palms, small middle phalanges of small fingers with a curvature, protruding abdomen. The head measured 32½ cm. in circumference; 10.6 cm. in length, and 9 cm. in width. Pulse was 140. There was a marked systolic murmur near the left sternal border of the heart. The *psychological examination* on admission showed a M.A. of 18 days, and an I.Q. of .86, (Kuhlmann-Binet, many mongoloids rate high soon after birth). The erythrocytes numbered 3,500,000 per cubic millimeter and the hemoglobin, 13 gms., (Fisher). The leukocytes numbered 11,400 per cubic millimeter with 51 per cent polymorphonuclear, 6 per cent transitionales, 42 per cent lymphocytes. The erythrocytes showed slight achromia, slight to moderate anisocytosis, rare target cells, and three normoblasts. The platelets were normal. The Hinton test was negative.

Course: The baby was a feeding problem from birth. He had respiratory distress. This became apparent especially with a superimposed respiratory infection. At 6 months of age the baby developed bronchopneumonia and became quite cyanotic. Thereafter, the liver was enlarged and felt 3 mm. below the right costal margin. The pulse and respiration remained quite rapid with suprasternal and subcostal retractions. An electrocardiogram

was done and read by Dr. Conger Williams as follows: "This is a sinus tachycardia, rate of 150; P-R interval is 0.12 seconds; T-1, T-3 low. Chest leads show mostly upright QRS in V-3 and V-5. This tracing is within normal limits for this age of the patient." In spite of treatment the baby developed acute right heart failure with enlarged liver, cyanosis and died on October 1, 1955 at the age of 8 months.



Case 22—ECG.

Autopsy Findings: The pericardium contained 70 cc. of clear, straw colored fluids. There was a thin, fibrinous infiltration over the pericardial surface. The heart was moderately large weighing 44 gms., (normal weight, 37 gms.). It measured 65 mm. in vertical diameter, 50 mm. in transverse diameter and 80 mm. in oblique diameter. The aortic valve measured 20 mm. in circumference and its cusps were normal. The aorta measured 20 mm. in circumference at the valve. The aorta was relatively narrow; the cusps showed moderate fenestration. The pulmonary valve measured 30 mm. in circumference, and the pulmonary artery, 30 mm. at the valve. The pulmonary cusps showed moderate fenestration and there was also adherence between the posterior and anterior cusps. The coronary arteries were normal and in normal position. The right atrium was large measuring 60 mm. in transverse diameter, and 20 mm. in sagittal diameter, (including the auricular appendage). The venae cavae and coronary veins

were normal. The left atrium measured 45 mm. in transverse diameter, 20 mm. in sagittal diameter, and received four pulmonary veins. There was a large defect of the lower part of the interatrial septum. The ventricle was divided by an incomplete septum which showed 5 mm. thickness and extended up for a distance of 27 mm. from the apex and left a large, round defect superiorly, measuring 25 mm. by 25 mm. The mitral and tricuspid orifices were not separated, and one large atrioventricular orifice was present which measured 70 mm. in circumference. The common artioventricular valve measured 65 mm. in circumference and was composed of four leaflets. Two of these were connected with the pulmonary conus and the one large one was located on the middle line of the common ventricle, (medial leaflet). The other laid in the left ventricle. The wall of ventricles measured 9 mm. in thickness. The foramen ovale measured 3 mm. in diameter. The circumference of the ductus arteriosus was 7 mm. Double common carotid arteries were observed on the left side. *Other findings at autopsy:* Hemorrhagic bronchopneumonia, (aspiration pneumonia); marked fatty degeneration of the liver; hypoplasia of adrenal and pituitary glands.

Diagnosis: Mongolism; ostium atrioventriculare commune; patent ductus arteriosus; dilatation of the pulmonary trunk; dilatation of the right atrium and right ventricle; underdeveloped pulmonary conus; malformation of left common carotid artery; patent foramen ovale.

IX. *Case 23.* M.D., white male, born August 12, 1955 at 8 months, 3 weeks by normal delivery. Birth weight was 4 pounds, 7 ounces. There was a venous thrombosis starting 3 cm. from the umbilicus and extending for 10 cm. Third pregnancy. Mother was 37 years of age at birth of patient. Her health was good in general during pregnancy. She has a rheumatic heart disease. The parents are of normal intelligence. The results of the first and second pregnancy were normal children, physically and mentally. The family history was negative on both sides. Mongolism was diagnosed at birth, and institutionalization was recommended. The baby was transferred to the Wrentham State School on August 31, 1955 at the age of nineteen days.

The *physical examination* showed a tiny, jaundiced infant who was 47 cm. tall and weighed 2270 gms. The umbilical stump was covered with purulent material. He showed typical mongoloid features with slanting palpebral fissures, flattened helices, a right

epicanthal fold, a protruding tongue, four finger (Simian) line on left hand, small nose with broad nasal bridge, hypotonic muscles, asymmetric head with occipital bulge. The anterior and posterior fontanelles were largely open. There was a marked systolic murmur along the left sternal border of the heart. The pulse was 136. The liver was felt 2 cm. below the right costal margin. The head circumference was 33 cm. The *psychological examination* revealed a M.A. of 1 week, and an I.Q. of .33. The erythrocytes numbered 4,700,000 per cubic millimeter, and the hemoglobin, 17.2 gms., (Fisher). The leukocytes numbered 7,400 with 41 per cent polymorphonuclear, 21 per cent monocytes, 38 per cent lymphocytes. The blood platelets were normal. The erythrocytes showed slight achromia, moderate anisocytosis and slight poikilocytosis.

Course: The baby took his feeding well, but showed very little weight gain. He had an infection of the umbilicus. At the age of six weeks, jaundice regressed, but his pallor was increased. He was treated by chloromycetin and iron. In spite of treatment, the child died at the age of 2 months, 10 days.

Autopsy Findings: The pericardium contained a few droplets of clear, straw colored fluids. The heart was very small weighing 15½ gms., (normal weight, 23 gms.). It measured 35 mm. in vertical diameter, 30 mm. in transverse and 38 mm. in oblique diameter. The aortic valve measured 23 mm. in circumference. The aorta 23 mm. in circumference at the valve. The aortic cusps were normal except for moderate fenestration. The coronary arteries were in normal position. The pulmonary valve measured 27 mm. in circumference, and the pulmonary artery measured 27 mm. in circumference at the valve. The both atria were large. The right atrium measured 40 mm. in transverse diameter, and 20 mm. in sagittal diameter, (including the auricular appendage). The venae cavae and coronary veins were normal. The atrial septum was intact. The foramen ovale was patent measuring 5 mm. in diameter. The left atrium measured 35 mm. in transverse diameter, and 20 mm. in sagittal diameter. The pulmonary veins enter in their usual position. The ventricle was divided by an incomplete septum which showed 3 mm. thickening and extended up for a distance of 21 mm. from the apex, and left a relatively small round defect superiorly, measuring 7 mm. by 7 mm. The wall of ventricles measured 6 mm. in thickness. The mitral orifice measured 35 mm. in circumference and the mitral valve meas-

ured 30 mm. in circumference, and was moderately thick. The tricuspid orifice measured 45 mm. in circumference. The tricuspid valve measured 40 mm. and showed moderate thickness. There were pinhead sized hematomas on the mitral and tricuspid valves. The mitral cusps were normal but the medial cusp of the tricuspid was thick and adherent to the ventricular septum at the level of the defect. *Other findings at autopsy:* The ductus arteriosus was small and measured 3 mm. in diameter. Scattered atelectasis of lungs; dry, pale, reddish bone marrow; Meckel's diverticulum; marked adhesion between dura and the skull; congenital malformation of occipital bone, (the occipital bone was composed of five different small sized bones).

Diagnosis: Mongolism; interventricular septal defect; patent ductus arteriosus; abnormality of the tricuspid valve; patent foramen ovale.

X. *Case 24.* J.L., white mongoloid female, born July 26, 1940 by premature and instrumental delivery at eight months, because of mother's toxemia, (pre-eclampsia). Birth weight, 2,013 gms. Fourth pregnancy. The prenatal course was complicated by several episodes of bleeding. Mother had been frightened during the third month of pregnancy by what she thought was a burglar. Mother and the baby were discharged from the hospital nine days after delivery, the infant then weighing 2,466 gms. Mother stated that her whole life had changed both physically and mentally after giving birth to this child, and she became very sensitive and nervous. The baby was examined at the Boston City Hospital and a diagnosis of "mongolism with congenital heart disease" was made. Dentition started at thirteen months of age. She walked at sixteen months and started to talk at twenty months of age. Parents are of normal intelligence. Mother was 34 years of age at birth of patient. The first pregnancy resulted in an induced delivery at 6½ months because of toxemia, (pre-eclampsia) of the mother. The baby lived eight hours. The result of the second pregnancy was a normal, intelligent girl who is now married and has a normal baby. The third pregnancy resulted in a miscarriage at three months, attributed to mother's lifting heavy pails of water. This miscarriage happened one year before our patient's birth. Mother stated that she was not properly cared for after the miscarriage.

The patient was admitted to the Wrentham State School at the age of two years, one month. At first she appeared homesick and shy for a week or two but, generally, became better adjusted.

The *physical examination* revealed a blond mongoloid girl who weighed 11 kgs. and was 74 cm. tall. She had dark blue eyes with heterochromic irises, epicanthal folds, small nose with broad nasal bridge, small, simple ears with very small ear canals and scarred drums, chapped chin and cheeks, mottled cyanosis of skin, harsh breath sounds, and moderate sinus arrhythmia. She had four finger (Simian) line across the palms, a large fissured tongue, slanting eyes, narrow palate, and scoliosis of the lower dorsal spine. The head measured 46 cm. in circumference, 11½ cm. in width, and 14½ cm. in length.

In 1949, the *fluoroscopy examination* was done by Dr. Elkin. His report was as follows: "Examination of the heart by PA film and fluoroscopy shows the heart to be enlarged, measuring 10.6 cm. in transverse diameter with transthoracic measurement of 19.6 cm. In the PA picture, the heart appears enlarged generally but at fluoroscopy the enlargement appeared to be chiefly in the region of the right ventricle as well as auricular enlargement most likely in the right auricle. The right hilar shadows appear prominent, apparently due to engorged vessels, although no hilar density could be demonstrated. Impression: The appearance is that of a shunt from left to right such as an interauricular septal defect." The *psychological examination* revealed a mental age of one year and an I.Q. of .50. A more recent test, (1952), revealed a mental age of 2 years, 8 months and an I.Q. of .22, (Stanford-Binet).

The erythrocytes numbered 4,500,000 per cubic millimeter and the hemoglobin 11.4 gms., (Fisher). The leukocytes numbered 4,100 per cubic millimeter with 76½ per cent polymorphonuclear, 18½ per cent lymphocytes, 5 per cent monocytes. The sedimentation rate was 56 mm. at the end of one hour. Total serum cholesterol was 152 mg. per cent. The x-ray examination showed a slight deviation of the trachea which suggested enlarged thyroid on the right side. The basal metabolic test showed a rate of plus 27 per cent. The Hinton test was negative.

Course: The child had a good disposition and was in good health, ate and slept well, was tidy with clothing and personal habits, played and got along well with other children. She attended sense training class and entertainments. She tried to be helpful in dressing other children. She had several attacks of upper respiratory infections with tonsillitis. The patient developed exophthalmus in 1955, and at the same time weight loss and diarrhea were noticed. Her weights were as follows: October.

36 kg.; November, 35 kg.; December 1955, 34 kg.; January 1956, 33 kg.; February, 31 kg. On March 12, 1956 the patient had pneumonia, and in spite of antibiotic treatment, temperature went up to 104°, pulse 105, respiration 68, she became cyanotic and died on March 17, 1956 at the age of 15 years, 7 months, and 20 days.

Autopsy Findings: The heart was enlarged, weighing 200 gms., (normal for this age, 130 gms.). The shape of the heart was normal. The measurements were 120 mm. in vertical diameter, 85 mm. in transverse diameter, 140 mm. in oblique diameter. The pericardium contained 10 cc. of clear, straw colored fluid. The aortic valve measured 50 mm., and the aorta measured 50 mm. in circumference at the valve and was mildly hypoplastic. The aortic cusps showed marked fenestration with a moderate thickening and slight adherence. The coronary arteries were in normal position, but the right coronary showed four orifices, one was large and the other three small. There were scattered arteriosclerotic plaques in the intima of aorta. The pulmonary valve measured 70 mm. in circumference, and the pulmonary artery measured 70 mm. in circumference at the valve. The cusps of the pulmonary artery showed fenestration. The right atrium was large measuring 130 mm. in transverse diameter, and 40 mm. in sagittal diameter, (including the auricular appendage). The venae cavae and coronary veins were normal. There were a few aberrant chordae in the right artium located between the foramen ovale and the auricle. The interatrial septum showed a large defect measuring 25 mm. by 25 mm. The tricuspid orifice measured 120 mm. in circumference. The tricuspid valve, measuring 115 mm. in circumference, was moderately thick and had rather thin chordae tendinae. The orifice of the coronary sinus was large measuring 10 mm. in diameter. The left atrium measured 90 mm. in transverse diameter, and 40 mm. in sagittal diameter. The pulmonary veins enter in their usual position. The mitral orifice was 90 mm. in circumference, and the mitral valve 80 mm. in circumference. The mitral cusps and chordae tendinae were markedly thickened. The interventricular septum was intact. The wall of the left ventricle measured 17 mm. in thickness with hypertrophic papillary muscles. The wall of the right ventricle measured 11 mm. in thickness, and was heavily trabeculated. There were numerous aberrant chordae tendinae in the right ventricle. The foramen ovale measured 7 mm. in diameter. The ductus arteriosus was represented by a ligamentous cord. *Other*

findings at autopsy: Extensive, partially hemorrhagic bilateral lobar pneumonia; empyema; hyperthyroidism; hypoplasia of adrenal and pituitary glands; uteral polyps; left ovarian cyst; marked induration of the liver.

Diagnosis: Mongolism; persistent ostium primum; marked cardiac hypertrophy; patent foramen ovale; hyperthyroidism. The findings about the pathology of thyroid will be published in detail.

XI. *Case 30.* M.R.W., white male, born August 4, 1955 five weeks before the expected date. Second pregnancy. The baby was born three days after the rupture of membranes. Mongolism and congenital heart malformation was diagnosed at birth. Baby was not cyanotic, but was in an incubator for five days and oxygen was administered because of the heart condition. Birth weight was five pounds, one ounce. Mother was 37 years of age at birth of patient. The baby was admitted to the Wrentham State School on December 11, 1956 at the age of one year, four months and seven days. Mother's health was good during pregnancy but an older brother of the patient was quite sick with a severe upper respiratory infection during second month of mother's pregnancy with this boy. There was no rash. Parents and older brother are healthy and of normal intelligence. Family history is negative. The *physical examination* revealed a mongoloid infant who weighed 3,600 gms. and was 71 cm. tall; he had a brachycephalic head with a circumference of 44.7 cm., length, 15 cm., and width, 12.5 cm.; epicanthal folds with slanting palpebral fissures were present; the eyes were light brown with heterochromia; the nose was small with a broad nasal bridge; the little fingers were curved with short middle phalanges; no four finger lines were present. He had a large tongue and marked muscular hypotonicity; short extremities, hands and fingers as well as feet and toes. The heart was not enlarged, but there was a loud systolic murmur at the third intercostal space at left border of sternum. The *psychological examination* showed a M.A. of 8.2 months and an I.Q. of .50, (Cattell Infant Scale).

The erythrocytes numbered 3,650,000 per cubic millimeter and the hemoglobin 10.7 gms., (Fisher). The leukocytes numbered 7,500 per cubic millimeter with 36 per cent of polymorphonuclear, 56 per cent lymphocytes, 8 per cent monocytes. Platelets were normal. Polymorphonuclears show shift to left.

Course: The infant was active, responded well to his surround-

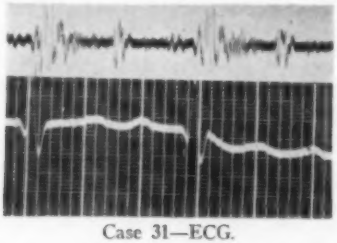
ings. At 16½ months of age he had an upper respiratory infection. Two months later he had bronchopneumonia with cyanosis and died at the age of one year, six months and fifteen days.

Autopsy Findings: The pericardium contained a few drops of straw colored fluid. Gross examination of the heart showed a large, strong right ventricle and right auricle. The heart was large weighing 90 gms., (normal weight for this age, 54 gms.). The vertical diameter was 80 mm., the transverse diameter 55 mm., and the oblique diameter, 70 mm. The aortic valve was 35 mm. in circumference and showed normal cusps. The aorta measured 30 mm. in circumference at the valve. The pulmonary valve measured 35 mm. in circumference, and the pulmonary artery measured 39 mm. in circumference at valve. The coronary arteries were normal and in normal position. The right atrium was large measuring 95 mm. in transverse diameter, and 35 mm. in sagittal diameter, (including the auricular appendage). The venae cavae were normal. The orifice of the coronary sinus was enlarged measuring 5 mm. in diameter. The left atrium measured 70 mm. in transverse diameter, and 35 mm. in sagittal diameter, and received four pulmonary veins in their normal position. The membraneous portion of the ventricular septum showed a sub-aortic round defect measuring 12 mm. by 10 mm. The aorta arose from both ventricles. The aortic cusps were normal. The pulmonary trunk was moderately dilated and also showed normal cusps. The mitral valve measured 50 mm. in circumference. The tricuspid valve measured 60 mm. in circumference and showed malformation of the valve; there was only two leaflets present. The wall of the right ventricle measured 11 mm. in thickness, and the wall of the left ventricle measured 9 mm. The ductus arteriosus was represented by a ligamentous cord. The foramen ovale was patent, measuring 8 mm. in diameter. *Other findings at autopsy:* Extensive bronchopneumonia; marked fatty degeneration of the liver; abnormal pulmonary fissure; congenital malformation of right vertebral artery.

Diagnosis: Mongolism; membraneous, sub-aortic interventricular septal defect; bi-ventricular origin of aorta; pulmonary dilatation; right cardiac hypertrophy, (Eisenmenger's complex).

XII. *Case 31.* P.D.L., white mongoloid male, born September 2, 1939 at full term. Third pregnancy. Mongolism was noticed shortly after birth. Mother was 26 years of age at birth of patient. She had an enlargement of thyroid, was very nervous

during pregnancy; vomited more during the first three months than in the other pregnancies. Parents and four siblings, two older, two younger, are of normal intelligence. Patient walked at 3½ years. Said a few words at the age of one year. On July 20, 1946 he was admitted to the Wrentham State School at the age of six years, ten months, because of the hardship he created for the family while at home. The *physical examination* on admission showed a well nourished mongoloid boy who weighed 36 pounds and was 39 inches tall. His head circumference was 17½ inches. He had left internal strabismus, slanting palpebral fissures, small nose with broad nasal bridge, short extremities, curved little fingers, short neck, large fissured tongue, pigeon breast, flaring ribs, small umbilical hernia, cyanotic mottling of the skin especially on the extremities, loud systolic murmur over the left sternal border, undescended testicles, four finger line (Simian line) in both palms. *Psychological examination* revealed a M.A. of 2 years, 8 months and an I.Q. of .39. The erythrocytes numbered 5,260,000 per cubic millimeter and the hemoglobin 15 gm. The leukocytes numbered 10,800 per cubic millimeter with 79 per cent polymorphonuclear, 21 per cent lymphocytes. The Hinton test was negative. The x-ray examination in 1948 was done by Dr. White. His report was as follows: "Examination of the chest shows enlargement of the heart, particularly on the left. There is prominence of the pulmonary conus and congestion in both lung fields. Findings are those of cardiac disease, probably congenital." X-ray examination of hands showed evidence of mongoloid deformities of the fifth fingers.



Case 31—ECG.

In 1946 an ECG was done and read by Dr. A. A. Luisada as follows: "Sinus rhythm. Prolonged P-L (Q-20). Severe right axis deviation. Intraventricular block. Phonocardiogram: Systolic murmur of medium intensity and of decreasing type at the apex. Very loud systolic murmur of a different type over the pulmonary

area; split second pulmonic sound. Third and auricular sounds. Interpretation: Intraventricular septal defect. Probably associated mild pulmonic stenosis."

In 1948, the fluoroscopic examination was done by Dr. Elkin. His report was as follows: "Examination of the heart by films and fluoroscopy showed it to be enlarged, measuring 11.9 cm. in transverse diameter with a transthoracic measurement of 20.5 cm. There is prominence in the region of the pulmonary artery as well as the right ventricle. There appeared to be no auricular enlargement. There is definite increase in the prominence of the hillar shadows bilaterally as well as in the pulmonary vascular markings. A definite hilar density could be demonstrated. The aortic arch appeared leftsided. There was a small, oblique pressure defect on the barium filled esophagus, coursing behind the esophagus and going upwards from left to right, very likely representing the crossing of the subclavian artery from an anomalous origin from the aorta. Impression: The appearance is that of a shunt from left to right as from an interauricular septal defect or a high interventricular septal defect. In addition there is an anomaly of the great vessels as manifested by the peculiar origin of the right subclavian artery." The patient had frequent upper respiratory infections. He developed severe pneumonia with dyspnea and marked cyanosis. In spite of treatment, he died on April 16, 1957.

Autopsy Findings: The heart was enlarged, weighing 410 gm. (normal weight, 130 gm.). The surface of the heart, especially over the anterior portion was covered with fibrinous-purulent exudate. The measurements were: 135 mm. in vertical diameter, 110 mm. in transverse diameter, 140 mm. in oblique diameter. The pericardium contained 25 cc. clear, straw color fluids. The aortic valve measured 50 mm. and the aorta measured 50 mm. in circumference at the valve. The aortic cusps were normal. The coronary arteries were normal and in normal position. The pulmonary valve measured 70 mm. and the pulmonary artery measured 70 mm. in circumference at the valve and was mildly dilated. The right atrium was large, measuring 155 mm. in transverse diameter, and 50 mm. in sagittal diameter (including the auricular appendage). The venae cavae and the coronary veins were normal. There was a large defect in the lower two-thirds of the interatrial septum, which combined with the interventricular septal defect. The orifice of the coronary sinus was large, meas-

uring 10 mm. in diameter. The left atrium measured 135 mm. in transverse diameter, and 30 mm. in sagittal diameter. The pulmonary veins enter in their usual position. The ventricles are divided by an incomplete septum, measuring 15 mm. in thickness which extends up for a distance of 65 mm. leaving a large round defect superiorly measuring 50 mm. x 50 mm. There was only one atrioventricular valve present because of the fusion of the tricuspid and the miltral cusps. The atrioventricular valve measured 150 mm. in circumference, and the atrioventricular orifice

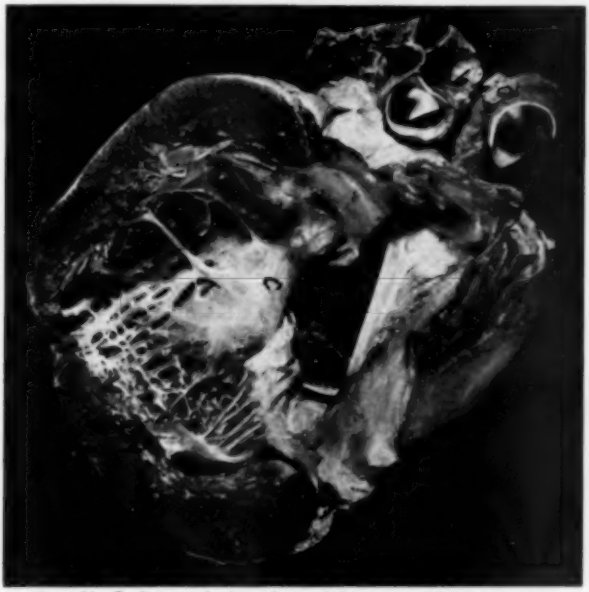


Fig. 2. Case 31. Left ventricular view. (a) aorta; (b) pulmonary artery; (c) remnant of ventricular septum.

160 mm. in circumference. The atrioventricular valve was composed of four leaflets. Two of them (lateral cusps) lay in the right ventricle and were attached to the papillary muscle of the right ventricle by chordae. A third leaflet (posterior cusp) was larger and laid on the anterior porton of the ventricular septum and attached partially on papillary muscle of the right ventricle, and papillary muscle of left ventricle by thick and short chordae. The fourth leaflet (anterior cusp) was also large and partly attached on the papillary muscle of left ventricle and on defective

septum and trabecular muscle in right ventricle. The functional edge of the atrioventricular valve and chordae tendineae were markedly thick. There were few thick aberrant chordae tendineae in the left ventricle. Both ventricles were heavily trabeculated throughout. The endocardium was markedly thick, especially on the left. The wall of the left ventricle measured 20 mm. in thickness, and the wall of the right ventricle 15 mm. The foramen ovale was patent and measured 10 mm. in diameter. The ductus arteriosus was represented by a ligamentous cord. The right subclavian artery arises as the fourth branch of the aorta and passes to the right behind the esophagus. *Other findings at autopsy:* Severe lobar pneumonia, empyema, acute pericarditis, congenital malformation of right middle lobe and fifth costa.

Diagnosis: Mongolism, ostium atrioventriculare commune, marked cardiac hypertrophy, patent foramen ovale. Abnormal origin of the right subclavian artery.

It is of special interest that this extremely severe malformation of the heart permitted this boy to live to the age of 17 years, 7 months and 15 days with very slight signs of circulatory distress.

DISCUSSION

Among the autopsies of thirty-one mongoloids we found twelve with congenital heart malformations, (38.8 per cent). Ostium atrioventriculare commune was observed in seven cases, (22.6 per cent of the mongoloids and 58.3 per cent of the heart malformations). An atrial septal defect occurred in two patients. A ventricular septal defect was observed in two cases. One case showed the Eisenmenger's complex. A patent ductus arteriosus was found in four patients. There was a high figure for hypertrophy of the right ventricle and hypoplasia of the left ventricle. Pulmonary stenosis was observed in one patient with ostium atrioventriculare commune. Dilatation of the pulmonary artery was seen in three patients. Abnormalities of the peripheral vessels occurred in three cases: in two the right subclavian artery arose as a branch of the descending aorta, instead of being a branch of the innominate artery. In the other case, there was a double carotid artery on the left.

The ostium atrioventriculare commune appears in two forms: a complete form and a partial form. Seven out of twelve autopsy cases of mongoloid children with congenital heart malformations showed the complete form of persistent ostium atrioventriculare commune, which is a defect of the lower part of the interatrial

TABLE I. DATA CONCERNING 30 MONGOLOIDS

CASE NO.	AUTOPSY NO.	SEX	I.Q.	AGE Yrs.	AGE Mos.	CAUSE OF DEATH	CONGENITAL CARDIAC DEFECT	OTHER DEFECTS
1	3/51	M.	.17	3	1	Bronchopneumonia.	Ventricular septal defect.	
2	1/52	F.	.26	1	3	Acute cardiac failure.	Ostium atrioventriculare commune; stenosis of pulmonary artery.	
3	10/52	F.	100		4	Bronchopneumonia.	Atrial septal defect; patent foramen ovale; left ventricular hypoplasia.	Small fissure of right pulmonary middle lobe.
4	11/52	M.	.38	8	½	Acute enterocolitis.	NONE	
5	13/52	M.	.14	16	1	Pulmonary tuberculosis. Epilepsy.	NONE	
6	14/52	M.	.67	1		Pneumonia; myocarditis.	Ostium atrioventriculare commune; patent ductus arteriosis; pulmonary artery dilatation.	
7	19/52	M.	.12	38	½	Pulmonary tuberculosis.	NONE	
8	20/52	M.	.21	6	3	Bacillary dysentery.	NONE	
9	21/52	M.	.35		7	Bronchopneumonia; myocarditis.	Ostium atrioventriculare commune; patent ductus arteriosus; pulmonary aneurysm.	
10	23/52	F.	.20	19	2	Endocarditis; thrombotica.	NONE	
11	8/53	M.	.21	21	9½	Pulmonary tuberculosis; amyloidosis; uremia.	NONE	
12	22/54	F.	.33		5½	Acute cardiac failure.	Ostium atrioventriculare commune; complete defect of ventricular septum; patent ductus arteriosus; patent foramen ovale.	
13	3/55	F.	.62	1	9	Aspiration pneumonia.	Ostium atrioventriculare commune; abnormal origin of right subclavian artery.	
14	10/55	M.	.33		4	Bacillary dysentery.	NONE	

TABLE I. DATA CONCERNING 30 MONGOLOIDS

CASE NO.	AUTOPSY NO.	SEX	I.Q.	AGE Yrs.	AGE Mos.	CAUSE OF DEATH	CONGENITAL CARDIAC DEFECT	OTHER DEFECTS
15	11/55	F.	.30	38	11	Pulmonary tuberculosis.	NONE	
16	12/55	M.	.20	58	9½	Bronchopneumonia.	NONE	
17	15/55	M.	.23	5	5	Bronchopneumonia.	NONE	
18	16/55	M.	.44	5	1	Bronchopneumonia.	NONE	
19	17/55	M.	.29	6	11	Bronchopneumonia.	NONE	
20	18/55	M.	.53	3	10	Bronchopneumonia.	NONE	
21	20/55	M.	.11	37	9½	Pulmonary tuberculosis; diffuse amyloidosis.	NONE	
22	32/55	M.	.86		8	Acute heart failure; aspiration pneumonia.	Ostium atrioventriculare commune; patent ductus arteriosus; patent foramen ovale; double carotid arteries; dilatation of pulmonary artery.	
23	33/55	M.	.33		2½	Bronchopneumonia.	Ventricular septal defect.	Malformation of occipital bone; Meckel's diverticulum.
24	10/56	F.	.22	15	7½	Lobar pneumonia, hyperthyroidism.	Atrial septal defect.	
25	14/56	F.	.20		11	Aspiration pneumonia.	NONE	
26	15/56	M.	.17	1	8½	Bronchopneumonia.	NONE	
27	17/56	M.	.09	29	6	Pneumonia; status epilepticus.	NONE	
28	25/56	F.	.19	47	11	Bronchopneumonia; intestinal tuberculosis.	NONE	Aplasia of right pulmonary middle lobe. Absence of right vertebral artery.
29	1/57	F.	.12	42	10½	Pulmonary tuberculosis.	Patent foramen ovale.	
30	4/57	F.	.50	1	6½	Bronchopneumonia.	Eisenmenger's complex.	Multiple fissures of lungs.
31	7/57	M.	.22	17	7½	Lobar pneumonia	Ostium atrioventriculare commune; patent foramen ovale; abnormal origin of right subclavian artery.	Malformation of right middle lobe, and fifth costa.

and the upper part of the interventricular septum, in which the heart is not divided in its four chambers. There is only a common atrioventricular orifice and valve present so that venous blood and arterial blood mix as they traverse the atrioventricular canal. A cleavage of the anterior mitral leaflet, and deformity of the tricuspid valve are constant features of the condition. In the partial form, usually there is a defect in the ventricular septum combined with atrial septal defect, but the tricuspid valve is properly formed. The mitral valve, on the other hand, shows a cleft in its anterior leaflet.

The clinical diagnosis of ostium atrioventriculare commune is difficult as murmurs are found in some cases only. Cyanosis occurs in superimposed pulmonary infections; the enlargement of the heart, the ECG is not significant. Also, Taussig stressed the difficulties to diagnose this condition without catheterization.

The heart develops from a straight tube which contains two series of endocardial cushions or endocardial thickenings. The proximal longitudinal cushions, A, B, and C, develop in the first stage of heart development and build the interventricular septum, part of the atrial septum, the tricuspid and mitral valves. The second row of cushions are the distal, horizontal cushions—an anterior, a posterior and two lateral—which develop into the cusps of aorta and pulmonary artery, and into the truncus septum. These parts of the heart develop in the second stage of heart development, (Tandler). The heart is formed when the embryo is about 2 mm. long in the early part of the fourth week, (Cunningham). At about the thirtieth day of intrauterine life, the cavity of the primitive atrium becomes divided into the right and left chamber by two septa: first, the septum primum, and at the thirty-fifth day of the intrauterine life by the septum secundum which grows downward between the atria and fuses shortly after birth with the primary septum closing the foramen ovale. In case of incomplete fusion, the foramen ovale remains open. Practically, also, at the thirty-fifth day of intrauterine life, the primitive ventricle becomes divided by a septum; septum inferius (ventricular septum), which grows upward from the lower part of the ventricle; its dorsal part increases more rapidly than its ventral portion and fuses with the dorsal part of the septum intermedium which arises from the posterior endocardial cushion. At the same time, the common atrioventricular canal becomes divided by the atrioventricular septum, but leaving the right and left margins of the canal intact producing the right and left atrioventricular orifices (Gray, Cunningham).

The second stage of heart development is not disturbed in mongolism when the distal four cushions develop into the cusps of the aorta and of the pulmonary artery and a part of the atrial septum. The inhibition of development in the first weeks of pregnancy supports Abbott's suggestion that the defect of development in mongoloids occurs early, and also fits well with the conception of Benda that at the time a growth stimulus is lacking which may normally be executed by the maternal pituitary gland.

Spitzer has claimed that the heart normally during its development performs a rotation of 180 degrees. If this rotation remains incomplete, heart malformations develop, (de-rotation). Evans has claimed that in mongoloids, the signs of the normal rotation are present which are missed in non-mongoloid heart malformations. He describes the rotation according to Keith's theory as rotation of the bulbus. Spitzer has discussed Keith's theory and proved its weakness. We would believe, too, that the rotation is a rotation of whole heart. However, we missed, in the mongoloid hearts, the signs of de-rotation, especially according to the location of the coronary arteries, which have been shown by Spitzer, and by Harris and Farber to be an excellent sign of the degree of rotation. This is a confirmation of Mautner's claim that heart malformations in mongoloids are mostly based on another mechanism as are malformations in non-mongoloids.

Abbott, the late student of heart malformations, said that the most interesting feature of the ostium atrioventriculare commune is the fact that it has been observed in mongoloids only. The fact that this malformation is seen almost exclusively in mongoloids proves that a special mechanism of development is active which must be related to mongolism. The explanation is probably that in the first weeks of pregnancy the development of the three proximal cushions is stopped. When development starts again, it does not start where it has been stopped, but according to the stage where it would be at this time in normal development. This means that the growth stimulus did not operate in the first stage of heart development in mongoloid children, but the second stage goes on normally.

The pathogenesis of mongolism is not clear. Benda claimed that in the early stage of intrauterine development, a growth stimulus is missing, probably normally produced by the maternal pituitary. Ingalls stated that mongolism is caused by any damage to the fetus in the first weeks of development which interferes with normal nutrition, especially with oxygen supply.

TABLE II. DATA CONCERNING THE MOTHERS

CASE NO.	AGE AT BIRTH OF CHILD	PHYSICAL CONDITION OF THE MOTHER DURING PREGNANCY	PREGNANCIES OF THE MOTHER
1	21	Good Health.	1st, our patient; 2nd and 3rd, normal.
2	33	Extremely nervous in 1½ months of pregnancy because of son's operation.	1st, normal; 2nd, our patient.
3	40	Vaginal bleeding at 2nd and 3rd month of pregnancy (threatened abortion). Became pregnant soon after miscarriage.	1st, premature twins, (hydramnion and toxemia), one died; 2nd, normal; 3rd, miscarriage, (threatened abortion); 4th, our patient.
4	35	Severe disease of siblings (cold).	1st and 2nd, normal; 3rd, our patient; 4th—8th normal.
5	18	Miscarriage shortly before pregnancy.	1st, miscarriage; 2nd, our patient.
6	40	Rheumatic heart disease; nervous.	1st and 2nd, normal; 3rd, twins, one died, other a mongoloid, living; 4th, our patient.
7	42	Severely depressed.	1st and 2nd, normal; 3rd, our patient.
8	37	Vaginal bleeding; two miscarriages prior to this pregnancy (abortion). Extremely disturbed.	1st and 2nd, miscarriages; 3rd, our patient.
9	27	Rheumatic heart disease; vaginal bleeding during first 2½ months.	1st, normal; 2nd, our patient.
10	37	Hypertension; asthma.	1st, 2nd, 3rd, normal; 4th, our patient.
11	35	Hypertension.	1st and 2nd, normal (one died of childhood disease); 3rd our patient.
12	27	Severe upper respiratory infection during 2nd month.	No other siblings.
13	36	Rheumatic heart disease.	1st and 2nd, normal; 3rd, our patient.
14	27	Became pregnant shortly after miscarriage. Contracted measles; disturbed.	1st normal; 2nd, miscarriage; 3rd, our patient.
15	38	Nervous; frightened by mad dog.	1st, 2nd, normal; 3rd, our patient.
16	39	Good health.	1st, 2nd and 3rd normal; 4th our patient.
17	21	Accident in 3rd month; disturbed.	1st, our patient; 2nd, normal.
18	42	Upset about pregnancy.	1st and 2nd, normal; 3rd, our patient.
19	39	Worried about financial situation.	1st, normal; 2nd, our patient.
20	44	Upper respiratory infection during first two months.	1st, 2nd, 3rd and 4th normal; 5th, our patient.
21	47	No history.	1st, 2nd, 3rd, normal; 4th, our patient.

TABLE II: DATA CONCERNING THE MOTHERS

NO. CASE	AGE AT BIRTH OF CHILD	PHYSICAL CONDITION OF THE MOTHER DURING PREGNANCY	PREGNANCIES OF THE MOTHER
22	32	Became pregnant shortly after miscarriage. Vaginal bleeding in early pregnancy (threatened abortion).	1st, normal; 2nd, miscarriage; 3rd, our patient.
23	37	In good health.	Two older, normal siblings; 3rd, our patient.
24	34	Became pregnant shortly after miscarriage. Pre-eclamptic.	1st (toxemia of mother), died after birth; 2nd, normal; 3rd, miscarriage; 4th, our patient.
25	40	Head injury with loss of consciousness in 2nd month.	Three older siblings, normal; 4th, our patient.
26	33	Severe upper respiratory infection at first month of pregnancy.	Three older siblings, normal; 4th, our patient.
27	42	In good health.	Seven older, one younger, normal siblings; one died of pneumonia; one miscarriage.
28	41	Psychotic.	Eight older siblings including twins. One of twins died; 9th, our patient.
29		No history.	No history.
30	37	Contracted severe upper respiratory infection during 2nd month.	1st, normal; 2nd, our patient.
31	26	Enlargement of thyroid; extremely nervous.	1st, 2nd, normal; 3rd, our patient; 4th and 5th, normal.

Development of malformations due to extrinsic damage to the fetus is a widely discussed chapter. Heart malformations have been connected with such damages, too. In animal experiments, Buechner produced such malformations by low oxygen supply, Wilson and Warkany by vitamin A low diet, Wolff by x-ray radiation. Heart malformations in children were attributed by Plummer to radiation after the Hiroshima blast. Since Gregg's first publication, a series of heart malformations were described in children whose mothers had rubella during early pregnancy; other diseases probably play the same role. On the other hand, a heredity tendency of heart malformations has been claimed, (Rösler, Campbell, McKeon, etc.). It could be that in children of such families early damage to the fetus will cause heart malformations in a higher percentage than in other children. Then too, other conditions in early pregnancy may have etiological importance for heart malformations. They have been found in diabetic mothers, (Hurwitz, Higgane). In addition, attempted abortion (Husler) or curettage (Mayerhofer) have been con-

nected with mongolism and may, therefore, also be responsible for the heart malformations. Development of heart malformations does not depend on the type of damage but on the time of the damage during pregnancy.

Most of the heart malformations in our mongoloid cases indicate that the development of the heart was arrested at the first stage of heart development which takes place in about the first seven weeks of pregnancy.

SUMMARY

1. Heart malformations in mongoloids have been discussed.
2. In thirty-one autopsies, twelve heart malformations were found.
3. A relatively large number (58.3 per cent) of heart malformations in mongoloids was ostium atrioventriculare commune.

REFERENCES

1. Abbott, M.: Atlas of Congenital Cardiac Disease. Am. Heart A., 1936.
2. Benda, C. E.: Mongolism and Cretinism. New York, 1946.
3. Buechner, F.: Exp. Entwicklungestoerungen Durch Sauerstoffmangel. Klin. Wchnschr., 24:137, 1946.
4. Campbell, M.: Congenital Heart Disease. Guy's Hosp. Gaz., 60:150, 1946.
5. Conte, W. R.; McCammon, C. S. and Amos, C.: Congenital Defects Following Rubella. Am. J. Dis. Child., 70:301, 1945.
6. Cunningham, J. H.: Text-Book of Anatomy. New York, 1937.
7. Evans, Ph. R.: Cardiac Anomalies in Mongolism. Brit. Heart J., 12:258, 1950.
8. Gray, H.: Anatomy of the Human Body. Philadelphia, 1936.
9. Gregg N. M.: Congenital Cataract Following German Measles in Mother. Tr. Ophth. Soc. Australia, 3:35, 1941. (Cited by Swan).
10. Harris, J. S. and Farber, S.: Transposition of the Great Cardiac Vessels, with Special Reference to the Phylogenetic Theory of Spitzer. Arch. Path., 28:427, 1939.
11. Hurwitz, D. and Higgens, N.: Diabetes in Pregnancy. New England J. Med., 247:305, 1952.
12. Husler: Handbuch Der Kinderheilkunde. Berlin, 1931.
13. Ingalls, Th. H.: Etiology of Mongolism. Am. J. Dis. Child., 165:147, 1947.
14. Ingalls, Th. H.; Curley, F. C. and Prindle, R. A.: Anoxia as Cause of Fetal Death or Congenital Defect in the Mouse. Am. J. Dis. Child., 80:34, 1950.
15. Kaiser, A. and Schmidt, F.: Die Herzfehlerbildung beim Mongolismus. Munchen. med. Wchnschr., 94:2167, 1952.
16. Keith, A.: Abstract of the Hunterian Lectures on the Evolution and Action of Certain Structures of the Heart. Lancet, Vol. 1, Nos. 9, 10, 11, 1904.
17. Mautner, H.: Congenital Heart Diseases in the Feebleminded. ARCH. PEDIAT., 55:546, 1951.
18. Mautner, H.: The Developmental Mechanics of Heart Malformations. ARCH. PEDIAT., 73:47, 1956.
19. Mayerhofer, E.: Die Mongoloide Idioite mit Ihrer Ursachlicher Beziehung zu Curettage-Abortus. Ann. Pediat., 154:57, 1939.
20. Plummer, G.: Anomalies Occuring in Children Exposed in Utero to the Atomic Bomb in Hiroshima. Pediatrics, 10:687, 1952.
21. Spitzer, A.: Ueber den Bauplan des Normalen und des Missbildeten Herzens. Versuch Einer Phylogenetishen Theorie. Virchows Arch. f. path. Anat., 243:81, 1923.
22. Tandler, J.: Anatomie des Herzens. Jena, 1913.
23. Taussig, H. B.: Congenital Malformations of the Heart. New York, 1947.
24. Warkany, J.: Congenital Malformations Induced by Maternal Dietary Deficiency. Pediatrics, 25:476, 1944.
25. Wilson, J. G. and Warkany, J.: Aortic Arch and Cardiac Anomalies in the Offspring of Vitamin A Deficient Rats. Am. J. Anat., 85:113, 1949.

THE HAIR SIGN AS A PROGNOSTICATOR OF ALLERGY

ROBERT COHEN, M.D.
Bakersfield, California.

In 1925,[1] Freund described a tuft of hair over the vertex which he considered a premonitory sign of the exudative diathesis. Finkelstein preferred to consider this a sign of a neuropathic individual. This phenomenon aroused my interest sufficiently to follow it over a period of twenty years. Time has shown that the so called exudative diathesis is none other than the allergic and eczematous conditions of today. The neuropathic individual now is considered to have the irritabilities and neurological manifestations that accompany the allergic status.

This hair sign is not mentioned in our standard textbooks today on pediatrics or even allergies. It is thought of sufficient importance to bring it up as a sign in dealing with suspected allergy cases.

Over a period of twenty years, I have noted this hair sign and questioned the mothers as to whether they have had difficulty in making their infant's hair stay down with comb and brush. Their replies were for the most part in the affirmative. Further investigation into the parents' history with regard to allergies or allergic backgrounds were rewarded in about 75 per cent of the cases. However, the hair sign would alert us for closer observation, for other signs of eczema which in many instances were obvious. This condition can be used as an omen of impending trouble. Because of this sign, a very gradual introduction of new food regimes may be instituted and the possible development of an eczematous rash was stressed to the parents. It must also be kept in mind that some cases of thyroid deficiencies have thin hair and some a tendency to stand up when combed.

Being aware of this premonitory hair sign over a period of twenty years has enabled me to adopt a carefully planned feeding schedule, starting the diet with a soybean preparation, and then slowly introducing the least allergenic foods. In this way, it was easy to minimize the eczematous reactions so prone in allergic infants.

The same premonitory hair sign has helped in the infant diarrhea problems. When such cases were under protein milk or skimmed milk regime and failed to respond within a reasonable period of

time, they would be switched to a soybean milk preparation and the response was astonishing, the stools rapidly becoming normal. This fact was always kept in mind when going through a diarrhea ward and the infants with hair standing up tuffy like were spotted. A careful, retrospective study of the allergy history of those cases was made, and if positive, they were placed on a soybean milk regime.

As a typical illustration of its usefulness, the following case is cited. A five month old Spanish baby boy was seen with a history of green watery stools, very dehydrated as shown by sunken fontanelles, sunken eyes and poor tissue turgor. He was placed on a protein milk regime. The tenth day of the child's illness showed the same type of stools without any improvement. The infant's hair failed to stay down when brushed and gave the premonitory sign which is considered suggestive of allergy. The family history was definitely non contributory for asthma, hay fever, sinus trouble, or any food sensitivity. Nonetheless, a soybean milk preparation was then given and within twelve hours the stools became normal and the infant was discharged within twenty-four hours. He gained two pounds within a ten day period thereafter.

Premonitory Hair Sign.

Because of my increasing success with such treatment based on this sign, it is worthy of consideration by others.

REFERENCES

1. Feer, E.: The Diagnosis of Children's Disease. New York. 1925.

DEPARTMENT OF ABSTRACTS

Conducted by
MICHAEL A. BRESCIA, M.D., NEW YORK

KARLIN, I. W.: STUTTERING. EVALUATION AND TREATMENT. (New York State Journal of Medicine, 56:3719, Dec. 1, 1956).

Stuttering is a disorganization of the continuous rhythmic flow of speech. Its onset is in early childhood, and it has a lifelong history. On the basis of clinical observations stuttering is viewed fundamentally as an organic disorder. The theory that stuttering is due primarily to a slower process of myelinization offers a satisfactory explanation of the known basic facts. Emotional and environmental factors play an important role in unfolding and perpetuating the disorder. Emphasis shoulde be on immediate treatment of the preschool child who shows early signs of stuttering.
AUTHOR'S SUMMARY.

CARTER, C. H. AND MALEY, M. C.: CHLORPROMAZINE THERAPY IN CHILDREN AT THE FLORIDA FARM COLONY. (American Journal Medical Sciences, 233:131, Feb. 1957).

Chlorpromazine was administered for a period of six to eight months to 153 mentally defective, pre-psychotic, epileptic or spastic children. Daily dosages ranged between 30 mg. and 600 mg. with most patients receiving 75 mg. or 150 mg. Results were rated excellent in eighty cases (52%), good in thirty-nine cases (25%), fair in eighteen cases (12%), and poor in sixteen (10%). Hostility, hypochondria, fear, and withdrawal were greatly reduced or eliminated in individual patients, and general disturbances of ward routine by violence or apprehension were eliminated. Destruction of property was reduced. Pre-psychotics and mental defectives were relieved of disturbing symptoms and enabled to respond to training and schooling. Chlorpromazine reduced pre- and post-seizure tension and dread of approaching seizures in epileptics. The drug produced a good degree of muscular relaxation and increase in muscular coordination in spastics. A combination of chlorpromazine and mephenesin, or chlorpromazine and glutamic acid, gave better relaxation of skeletal muscles than did either chlorpromazine or the muscle relaxants alone. The over-all atmosphere of the institution was significantly improved. Conduct of programs was facilitated for the staff. AUTHORS' SUMMARY.

FARQUHAR, J. W.: THE POSSIBLE INFLUENCE OF HYPERADRENOCORTICISM ON THE FOETUS OF THE DIABETIC WOMAN. (Archives of Disease in Childhood, 31:483, Dec. 1956).

The foetus of the diabetic woman is oversized in length and weight, retains excessive fluid, shows hyperplasia of pancreatic beta cells and possible enlargement of the adrenal cortices. An attempt has been made to find a common cause for these, for the neonatal morbidity and for the maternal metabolic disorder. The possible growth-promoting, fluid-retaining and diabetogenic influences have been reviewed. Foetal hyperadrenocorticism is suggested by a number of morphological features. This in turn may stimulate the increased secretion of growth-promoting insulin. The absolute eosinophil count of the peripheral blood was determined at birth, 3 hours and 6 hours later, and daily for 10 days in thirty-two normal infants and in seventeen babies born to diabetic women. The range of birth levels in the two groups was very similar and was high. Maternal production of cortisone-like substances is high even in normal pregnancy. This would suggest that the eosinophil mechanism is inactive or incomplete before delivery. There was a consistently greater decrease in eosinophils in the abnormal group in the first 6 hours. The different routes of delivery may have accounted for this in part. The eosinophil behavior of the abnormal group was also more consistent from the second day and was characterized by a rise to about the end of the first week. This pattern suggested a greater response to stress the first 24-48 hours followed by a period of decreasing activity. The urinary excretion of acid-stable formaldehydrogenic steroids was employed as an index of adrenocortical activity. The infants of diabetic women showed a higher A.S.F.S. excretion than those of the controls on the first day, but this difference was lost by the third day. The 17-ketosteroid excretion of the two groups was similar. Infants of the abnormal group passed a greater volume of urine in the first 48 hours than the control infants. Their higher excretion of A.S.F.S., however, could not be explained on this basis. No relationship was found to exist between the hormone excretion values and neonatal behavior. The investigation was limited by the methods used to assess adrenocortical activity and by the equally great difficulty of obtaining satisfactory control groups. Further studies have been undertaken which may shed further light upon the possible influence of hyperadrenocorticism upon the development of the foetus of the diabetic mother. AUTHOR'S SUMMARY.

VARADI, S. AND HURWORTH, E.: HEINZ-BODY ANEMIA IN THE NEWBORN. (British Medical Journal, 5014:315, Feb. 9, 1957).

Two cases of spontaneous Heinz-body anemia are described. One, a full term twin of normal birth weight, had both hemolytic anemia and jaundice. The other, a premature infant, had typical hematological findings but no jaundice. Both responded well to a single transfusion. In the differential diagnosis of jaundice in the newborn, Heinz-body anemia should always be considered, especially if jaundice—not obstructive in character—develops after the first day of life, the Coombs test is negative, and no immune antibodies are present in the maternal serum. It would also appear that Heinz-body anemia can be present without clinical jaundice: if marked anemia develops in the premature infant during the second or third week and contracted irregularly crenated erythrocytes are found in the smears, Heinz-bodies should be looked for. It is likely that if these hematological criteria are systematically sought, more cases of this type of hemolytic anemia will be recognized in the future. Heinz-body anemia of the newborn is not only of theoretical but also of considerable practical importance as transfusions performed in time might save life.

AUTHORS' SUMMARY.

CHOREMIS, C.; PAPADATOS, C.; GARGOULAS, A. AND DROSOS, C.: INTRATHECAL HYDROCORTISONE IN THE TREATMENT OF TUBERCULOUS MENINGITIS. (Journal of Pediatrics, 50:138, Feb. 1957).

In this report a group of patients with tuberculous meningitis, treated by standard methods of therapy, is compared with a group of patients with the same disease who received in addition cortisone intramuscularly, and with a third group in which hydrocortisone was administered intramuscularly and/or intrathecally. We believe that I.M. cortisone has a beneficial effect on the clinical course and laboratory findings. Though we do not have enough evidence to support the claim that I.T. hydrocortisone prevents the occurrence of neurological complications, we nevertheless think that this mode of therapy is superior to all others, and that astonishing results occur in apparently hopeless situations. In two of our cases an early block in the circulation of the C.S.F. was relieved by cisternal infusions of hydrocortisone. The rapid clinical and laboratory improvement in some cases of patients with T.B.M. receiving I.T. hydrocortisone warrants a more extensive trial of its use.

AUTHORS' SUMMARY.

GELFAND, H. M.; FOX, J. P. and LE BLANC, D. R.: OBSERVATIONS ON NATURAL POLIOVIRUS INFECTIONS IN IMMUNIZED CHILDREN. (American Journal Public Health, 47:421, April 1957).

Some three hundred incompletely immune members of one hundred and eighteen Louisiana households were given a two-inoculation primary course of Salk vaccine. The serologic response at one month after the second inoculation has been determined by measuring the ability of the sera to inhibit the cytopathic effect of poliovirus. Overall, the three components of the vaccine were not equally antigenic, the Type 3 component being the least so. The de novo response to a particular type of antigen was enhanced materially by the preexistence of heterotypic antibody. In the first seven months following vaccination, child members of these households were closely observed for alimentary infections, a total of forty household episodes having been detected. These 1956 observations (made after vaccination) on the frequency of household episodes of infection, the extent of intrahousehold spread of virus, and the frequency, duration, and amount of virus excretion in the feces have been compared with similar observations made in the same study group in the period prior to vaccination. This comparison has led to the conclusion that two doses of Salk vaccine did not materially influence the frequency or duration of alimentary infection or the amount of virus excreted in the feces. The prediction is made that a third booster inoculation also will fail to influence enteric infection and that, as a corollary, extended use of killed-virus vaccines will not result in the gradual elimination of poliovirus from vaccinated areas. AUTHORS' SUMMARY.

BEACH, W. B.: PSYCHOSIS IN CHILDHOOD. (Northwest Medicine, 56:438, April 1957).

Schizophrenic reaction, the most important psychosis of childhood, is the end result of personality disorder brought about by stress which overrides the barriers possessed by the individual child. It is treatable. Therapy must be based on understanding of the factors in its origin and must include the entire family unit. Parents must be led to understand themselves in their emotional relationship to the child. The condition of children with actual mental retardation or brain damage may be much improved if the factors capable of producing schizophrenic reactions are abated.
AUTHORS' SUMMARY.

BAIN, H. W.; BOWDEN, D. H.; CHUTE, A. L.; JACKSON, S. H.; SASS-KORTSAK, A. and WALKER, N. F.: GALACTOSEMIA. (Canadian Medical Association Journal, 76:278, Feb. 15, 1957).

Galactosemia is a congenital, familial inborn error of metabolism characterized by severe malnutrition, hepatomegaly, presence of reducing substance (galactose) in urine, and perhaps, jaundice, cataracts, and mental retardation. Cases are frequently missed because of failure to consider the diagnosis in all cases of jaundice and dystrophy in young infants, and failure to test urine for reducing substance while the patient is receiving milk feedings. It is felt that the various manifestations are best explained by direct toxic action of galactose, and a theory as to the pathogenesis of liver damage is presented. All signs and symptoms, perhaps, with the exception of cataracts and mental retardation, are reversed when milk products are removed from the diet. Results of various laboratory tests, especially those of liver function, are presented. The genetic aspects are discussed and detailed pathology of two cases is presented. It is suggested that undiagnosed cases will be found in a review of case histories of autopsy cases of "cirrhosis" in early infancy. AUTHORS' SUMMARY.

CHALMERS, T. C.; IBER, F. L. AND UZMAN, L. L.: HEPATOLENTICULAR DEGENERATION (WILSON'S DISEASE) AS A FORM OF IDIOPATHIC CIRRHOSIS. (New England Journal of Medicine, 256:235, Feb. 7, 1957).

Five cases of cirrhosis of the liver associated with hepatolenticular degeneration without significant neurologic findings are reported in adolescents. Clinically, the defect might appear first as an asymptomatic aminoaciduria; secondly, as cirrhosis, chronic hepatitis, or Banti's syndrome ("abdominal" Wilson's Disease); thirdly, as typical hepatolenticular degeneration; fourthly, as the predominantly neurologic pseudosclerosis. The application of the biochemical abnormalities of this disease (aminoaciduria, peptiduria, hypouricemia, increased tissue copper, cupruria, and decreased serum ceruloplasmin content) to its diagnosis in the laboratory is discussed in detail. It is suggested that this "abdominal" form of Wilson's Disease may be a relatively common and previously overlooked cause of idiopathic cirrhosis in children and young adults. M.A.B.

AS THEY FIND IT–

"PRACTICAL"

The very concise and brief clinical reports that appear in the ARCHIVES each month have been very helpful to our readers, *who have found more practical suggestions in them than in most of the long and drawn out scientific articles that appear in other journals.*

We quote from a number of letters received from subscribers:

"I want to tell you that after subscribing for about 16 years, *I get more practical pediatrics from your journal than from either of the other pediatric journals to which I subscribe.*"

"Because it is *concise and progressive,* I consider ARCHIVES OF PEDIATRICS my favorite among pediatric journals. I always look forward with pleasure to the receipt of each number and *glean much of value from it.*"

"Only one other pediatric journal is now on file and I feel that *yours will be of more practical value to the general practitioners.*"

Archives of Pediatrics

(Names on Application)

The Vacant Chair

FOR ALL COMMON FORMS OF **DIARRHEA**

New 4-way check . . .

POMALIN *Liquid*

TRADEMARK

DEMULCENT · ANTI-INFECTIVE · ANTIDIARRHEAL

. . . effective against both specific and nonspecific diarrheas
. . . palatable oral suspension . . . well tolerated

FORMULA: Each 15 cc. (tablespoonful) contains:

Sulfaguanidine	2 Gm.
Pectin	225 mg.
Kaolin	3 Gm.
Opium tincture (equivalent to 2 cc. paregoric)	0.08 cc.

DOSAGE: ***Adults:*** Initially 1 or 2 tablespoonfuls from 4 to 6 times daily, or 1 or 2 teaspoonfuls after each loose bowel movement; reduce dosage as diarrhea subsides.

Children: ½ teaspoonful (=2.5 cc.) per 15 lb. of body weight every 4 hours day and night until 5 stools daily, then every 8 hours for 3 days.

Bottles of 16 fl. oz.

EXEMPT NARCOTIC. AVAILABLE ON PRESCRIPTION ONLY.

Winthrop LABORATORIES · NEW YORK 18, N. Y.